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Rare diseases

A bibliography on health care and daily life

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1 INTRODUCTION

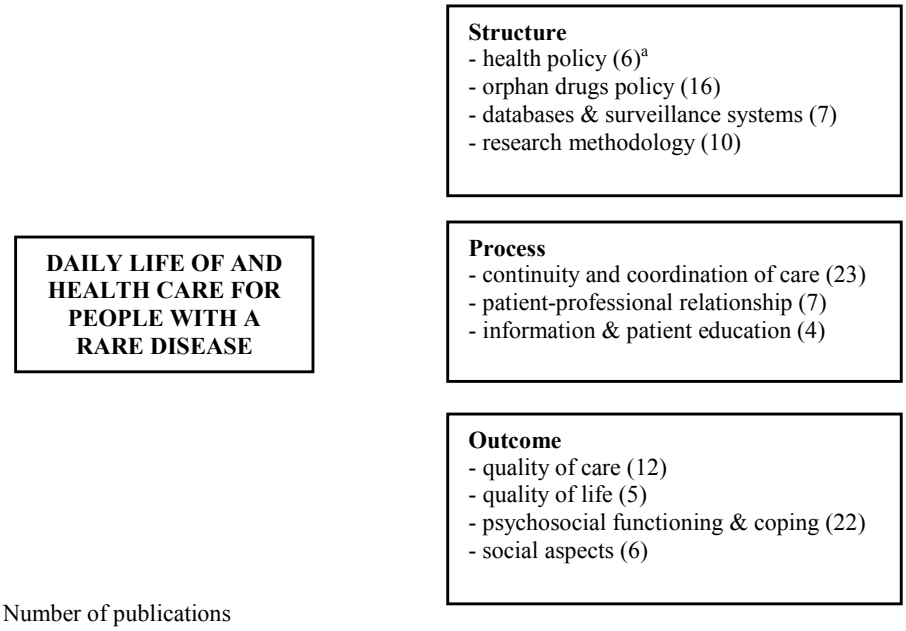
This bibliography has been designed to give an overview of research done and initiatives undertaken to improve daily life and health care in Europe for people with rare diseases. The development of drugs and treatment for specific rare diseases is growing and it is very important, but the 'human' perspective on rare diseases does not yet seem to have a place in the field of research. Rare diseases are defined as life threatening or chronic and progressive disorders with a prevalence of less than 5 in 10,000 persons (Orphanet, 2002; EC, 1999; RGO, 1998). Based on this European definition, 5,000 to 7,000 rare diseases can be distinguished. They are almost always debilitating and seriously harm the quality of life of the patients and the people near to them.

"Daily life of and health care for people with a rare disease" is a topic that concerns many fields. The purpose of this bibliography is beyond aetiology, medical treatment and other medical aspects of rare diseases. The topics of the bibliography are pointed out in Figure 1. The literature has been scanned on the basis of abstracts from selected publications. Subsequently, all topics were divided into different headings. Most documents are directly or indirectly related to improving the life of and care for people with rare diseases.

This bibliography is part of a larger project performed by NIVEL. The project consisted, apart from the bibliography, of empirical research on the specific problems and needs experienced by people with rare chronic diseases in health care and daily life (Van Nispen et al., 2003). This empirical study was funded by the Steering Committee Orphan Drugs[1], whereas the bibliography was financed by NIVEL.

[1] The Committee was appointed by the Dutch Minister of Health, Welfare and Sports in 2001, to encourage the development of Orphan Drugs and to improve the situation for patients with rare diseases.

Figure 1 Topics in the bibliography concerning people with rare diseases



[a] Number of publications

1.1 Search methods and selection criteria

1.1.1 Databases

Searches were conducted in medical, psychological, nursing and sociological databases, and in databases of a more general character. The reason to search through different databases was to find (research) documents on the subject of daily life of and health care for people with rare diseases that are beyond the medical aspects of the disease, like symptoms, medication or surgical treatment. Subjects of interest were, for example, quality of care, quality of life, psychosocial aspects of the disease and health policy (see topics in Figure 1). People suffering from rare diseases often consult health care professionals in several disciplines, such as physicians, psychologists, nurses and allied health care professionals. That is why databases of different disciplines had to be consulted. For instance, in a database like Psychinfo references can be found to literature about coping and quality of life of the chronically ill; a sociological database was selected in order to find literature about the position of chronically ill patients with rare diseases in society and

about policy on rare diseases and orphan drug production. Some general databases have been screened, because they cover a wide range of subjects. The database "Embase" was not selected, because a test search in this database resulted in only a large amount of purely medical publications. The following databases were consulted:

1 Pubmed, a database which includes Medline, is the most frequently used medical database. The database is set up by the National Library of Medicine of the United States and contains more than 11 million references to articles of approximately 4,300 scientific medical journals in many different languages.
2 Psychinfo, a database containing references to articles of approximately 1,300 international journals, books, chapters and dissertations in different languages, in the discipline of psychology and psychological aspects in medicine, psychiatry and nursing.
3. Catalogue of the Netherlands Institute for Health Services Research (NIVEL), which contains references to books, reports and journal articles, mostly in Dutch or English.
4. Cinahl, a database which contains references to English journal articles in the field of nursing and allied health care.
5. Sociological Abstracts, an international database containing references to articles of approximately 2,500 journals in the disciplines of sociology, social psychology, political sciences and environmental studies.
6. Social Science Citation Index, a general international database containing references to journal articles of all social scientific disciplines.
7. Science Direct, a database which contains the literature of all periodicals published by Elsevier.
8. Picarta is one of the largest databases available in the Netherlands, containing books, articles, abstracts, letters and internet-services. It includes a broad range of subjects. The catalogue gives access to databases, full text journals and to more than 400 Dutch libraries. Also, online contents of 12,000 journals are included.
9. 'The informed general practitioner' (De geïnformeerde huisarts), a database of Dutch literature with references to primary health care articles.
10. Catalogue of the 'Erfocentre', a Dutch organisation which informs the general public about heredity and health. The catalogue contains references to Dutch and English literature.

abstracts were screened on the basis of (the inclusion and) eight exclusion criteria:

1. The disease which is discussed is not rare, according to the Orphanet-index, i.e. an on-line encyclopaedia on 3,500 rare diseases (Orphanet, 2003; www.orpha.net).
2. Exclusively a description of the rare disease itself, like symptoms or diagnosis.
3. Exclusively a description of treatment options for a specific rare disease.
4. Exclusively a description of costs and economic aspects of treatment for a specific rare disease.
5. Exclusively a description of issues concerning genetic screening of a specific rare hereditary disease.
6. Health care policy, which is not relevant to the European situation.
7. Other criteria, like genetic structure underlying a disease or epidemiology.
8. Journalistic articles.

1.1.4 Search results

Table 1 shows the results, including the total number of titles found (N=3,226 'hits') and the number of excluded (N=3,106) and selected documents (N=135). Apart from the literature found in the different databases, there are two reports, sent by a patient organisation that was aware of the activities to compose a bibliography. After this contact, three more reports were obtained from the same organisation. The same happened when a report on orphan drugs was sent to the NIVEL-library by a health insurer: one more report was obtained. Two reports were sent to the library by others. After the searches were finished in August 2002, the second author came across six very recent publications, which were found coincidentally, while screening journals for the NIVEL-library. These magazines were a Dutch journal for medical science (Nederlands Tijdschrift voor de Geneeskunde), Patient Education and Counselling, Joint Commission Journal on Quality and Safety and the Journal of Advanced Nursing. These publications were considered to be very relevant and could not be excluded from the bibliography. The number of finally selected documents (N=118) is based on the total number of relevant documents (N=135) minus those which were found for a second time in another database (N=17). Finally, six publications that seemed relevant to include in the bibliography were not available in the Netherlands or outside the Netherlands for unknown reasons.

Table 1 Search results

Database name	Total number of hits	Total number of excluded documents	Number of relevant titles
Pubmed	1,454	1,396	58
Psychinfo	123	117	6
Catalogue NIVEL	131	122	9
Cinahl	521	508	13
Sociological abstracts	19	15	4
Social Sciences CI	75	65	10
Science direct	169	165	4
Picarta	575	563	12
The informed GP	148	147	1
Erfocentre Library	11	8	3
Not in database	15	-	15
Total	3,241	3,106	[a] 135

[a] Seventeen relevant publications were double. The total amount of relevant publications is 118.

Table 2 shows the number of excluded documents for each database and for each exclusion criterion (see §1.1.3). Documents that were found in more than one database were all counted. It is therefore not clear how many doubles were excluded.

Table 2 Number of excluded documents per database and per criterion

Exclusion criteria	1	2	3	4	5	6	7	8	Total
Pubmed	834	214	255	6	12	25	40	10	1,396
Psychinfo	70	31	6	-	-	-	10	-	117
Catalogue Nivel	114	-	1	-	-	-	2	5	122
Cinahl	263	94	86	2	3	24	17	19	508
Sociological abstracts	12	-	-	-	-	2	1	-	15
Social Sciences CI	19	4	2	-	3	2	31	4	65
Science direct	154	1	-	-	1	-	9	-	165
Picarta	13	224	75	5	38	59	86	63	563
The informed GP	139	4	2	-	-	-	1	1	147
Erfocentre Library	-	2	-	-	-	-	3	3	8
Total	1,618	574	427	13	57	112	200	105	3,106

Table 3 shows the number of relevant documents, divided into different periods of publication. Especially in the second half of the nineteen nineties,

many documents were found (42%). Also in the last 2,5 years (2001 - June 2003), the authors came across relatively many publications (26%).

Table 3 Number of relevant documents divided into publication period

Publication period	Number of documents	%
1976-1980	2	1.7
1981-1985	6	5.1
1986-1990	11	9.3
1991-1995	19	16.1
1996-2000	49	41.5
2001-2003	31	26.3
Total	118	100.0

Table 4 shows the number of relevant documents, divided into the languages of publication. Most documents are written in English (68%). Also, relatively many Dutch publications were found (22%), probably due to the databases that were selected.

Table 4 Number of relevant documents divided into languages

Language	Number of documents	%
English	80	67.8
Dutch	26	22.0
German	4	3.4
French	2	1.7
Italian	3	2.5
Norwegian	2	1.7
Swedish	2	1.7
Total	118	100.0

Table 5 shows the number of relevant documents divided into country of origin. Most documents that were selected are written by authors from the United States (N=36) and The Netherlands (N=31). Relatively more relevant documents are written by European authors (N=73, 62%) than by non-Europeans (N=45, 38%).

Table 5 Number of relevant documents divided into country of origin and publication type

Country	[a]Research	Other
Europe:	[b]1	1
Austria	-	1
Belgium	1	-
Finland	1	-
France	1	2
Germany	2	3
Italy	1	5
Norway	2	4
The Netherlands	14	17
United Kingdom	7	5
Spain	1	-
Sweden	2	2
Outside Europe:		
Australia	-	3
Canada	2	4
USA	17	19
Total	52	66

[a] Journal articles (N=41) and reports (N=11); [b] Cooperation between authors from Spain, Sweden, France and the United Kingdom.

One document is published by the European Community and one by a cooperation between authors of four different European countries. Furthermore, publication type was generally divided into research articles (N=41) and research reports (N=11) and other publications (N=66; general information, reviews, etc.).

1.2 Structure of health care

1.2.1 Health policy

Documents on health policy concerning rare diseases in different European countries were explored. Of specific interest were documents on actions undertaken, mainly in the European Union, to improve the situation for people with rare diseases. Six relevant articles and reports were found in the databases. One document contained an action programme by the European Community concerning research and management of rare diseases in the years 1999 to 2003. Documents on health policy outside Europe were excluded, but in some articles American policy was described together with the European one. Also, an American article on policy in managed care for patients suffering cystic fibrosis was included. Managed care is slowly

gaining territory in Europe, for example in Switzerland, and could be of particular interest to some readers (Colombo, 2001; European Observatory on Health Care Systems, 2000; Robinson, 1998).

Although articles on genetic screening for specific rare diseases were left out of the bibliography, one general publication on the ethical, legal and social problems concerning genetic screening for rare diseases was included. Furthermore, priorities for research regarding rare diseases are outlined and recommendations are given to improve information supply, research policy and development of orphan drugs.

☞ Go to page 23 to find abstracts on health policy.

The six publications include a report, a book chapter and four journal articles with general information or opinions. The communal action program is written in English, Dutch and other languages. Three publications are written in English and two in Italian.

1.2.2 Orphan drugs policy

The topic of orphan drugs policy has been described in 16 publications. Orphan drugs are specifically developed to treat rare diseases. Due to high development costs and low market profits (because of a low demand), developing these drugs is not very attractive for the pharmaceutical industry. This unfavourable situation changed by the American, and later the European, “Orphan Drug Act”. This Act lays down the conditions for attribution of the orphan drug status; it also draws up attractive proposals for pharmaceutical industries in order to improve the development of these drugs. The Orphan Drug Act was officially implemented in all countries of the European Union in the year 2000.

Some authors give background information on the Orphan Drug Act and regulations, but also recommendations for policy and research on these specific treatments are given. In other articles, the policy of the European Union regarding research and development of orphan drugs is described. Furthermore, recommendations concerning coordination and priority setting in the field of orphan drugs are outlined. In two reports, recommendations are given for the evaluation procedure for including orphan drugs in the social insurance package in The Netherlands. Also, the Dutch policy on orphan drugs in general is addressed in several publications. The public policies of six countries - Canada, France, Japan, Sweden, the United Kingdom, and the United States - affecting the development and marketing

of pharmaceuticals for rare diseases are compared in a research article. Finally, a second research article compares changes in orphan drug production before and after the American Orphan Drug Act of 1983. The recommendations given in this article might be useful for European research into the law's costs and benefits, but also for patients and the general public.

☞ Go to page 26 to find abstracts on orphan drugs policy.

The sixteen publications include five reports, two research articles, nine journal articles with general information or opinions and a Dutch governmental publication. Five publications are written in English, ten in Dutch and one in French.

1.2.3 Databases and surveillance systems

Only a few publications address databases and surveillance systems. Nevertheless, three articles describe the topic of surveillance of rare diseases in children. An example is the Dutch Paediatric Surveillance System. The goal of this system is to gain insight into the prevalence of rare and new diseases and to promote scientific research. Another initiative is the Australian Paediatric Surveillance Unit, aiming to collect information on the epidemiology of rare or uncommon childhood conditions. Also, a Spanish surveillance is described in a research article on five years of experience with a particular severe rare disease in adults (alpha-I-antitrypsin deficiency). Another initiative to easily find case histories of a specific rare disease on the internet is explored. Finally, a physician's guide to rare diseases with ready access to signs and symptoms on 700 rare diseases was found.

☞ Go to page 35 to find abstracts on databases and surveillance systems.

The seven publications include a reference work, five journal articles with general information or opinions and a research article. Six publications are written in English and one in Dutch.

1.2.4 Research methodology

Ten articles on research methodology were included in the bibliography. Two articles discuss how the internet can be useful to collect data on people with rare diseases. Also, in three other publications, other ways to register patients with rare diseases are suggested. One study explores how cooperative clinical trial groups can offer opportunities to do epidemiological research. Also, four articles discuss the impossibility of

randomised clinical trial research for rare disease treatment and suggest a different clinical trial design.

☞ Go to page 39 to find abstracts on research methodology.

From ten publications, six are research articles, three are journal articles on general information and opinions and one is a review article. All publications were written in English.

1.3 Health care process

1.3.1 Continuity and coordination of care

Twenty-three publications addressing continuity and coordination of care for people with rare diseases were selected in the bibliography. The choice to assign ten publications on health care centres or programmes, which is a typical "structure" topic, under the heading "health care processes" might be considered arbitrary. Nevertheless, the authors paid more attention to the process aspects than to the structure aspects of these centres. For example, one article gives a model of a research centre, which is focussed on the understanding of possible causes, prevention and treatment of rare diseases. Different definitions were considered to address the subject of continuity and coordination of care, such as concentration of knowledge, cooperation between hospitals, the possibility to easily share information, multi-disciplinary goals or meeting facilities for patients. In one of the articles, the authors describe their experience in developing a multi-disciplinary paediatric multiple sclerosis clinic, addressing both health and developmental needs of these children. Other authors give information on the way they concentrated their expertise in some Dutch liver centres. Similarly, three Swedish haemophilia centres have designed a programme to guarantee patients comparable and optimal care.

The topic of coordination and continuity of care has further been elaborated in thirteen other publications, in which, for example, the need for international cooperation in the care for children with kidney disease is considered. Although case studies were excluded from the bibliography, one was included, because it gave an example of the role of health-team members in assisting a child and family to manage the illness. A Dutch report recommends cooperation between different health care services to solve problems after diagnosis of a rare disease. One research article considers the role of the general practitioner, allowing the GP a more

prominent place in the care for people with rare diseases, even when the disease requires treatment by (super) specialists. One research article examines the role of health visitors (nurses) in diagnosing and managing acute childhood illness. A third research article investigates management of a rare disease in specialist and non-specialist clinic settings; centralisation of expertise and patient management in multi-disciplinary settings is recommended. Similar results were found in another study, e.g. more rare tumours were diagnosed in the specialised institute than in community hospitals. Furthermore, in four different reports the activities of the Dutch Neuromuscular Diseases Association were studied.

☞ Go to page 46 to find abstracts on continuity and coordination of care.

The total of 23 publications include five reports, three research articles, thirteen journal articles with general information or opinions and two articles in which research on a specific disease is partly reviewed. Ten publications are written in English, seven in Dutch, two in German, two in Swedish, one in Norwegian and one in Italian.

1.3.2 Relationship between patient and health care professional

In seven publications the topic of patient-professional relations are addressed. One research article investigates needs, experiences and expectations of primary care reported by patients. Furthermore, a process for delivering bad news to families of children with rare diseases has been investigated. One research article reports on parents' experiences of receiving a rare diagnosis of their children. Finally, one review article describes a literature search performed to investigate the relationship between GPs and patients with rare, serious chronic diseases. One study by the same authors mentions a shortage of expertise on rare diseases in family practise.

☞ Go to page 63 to find abstracts on patient-professional relationship.

The seven publications include two research articles, two review articles, two journal articles with general information or opinions and a report. Four publications are written in English, two in Dutch and one in German.

1.3.3 Information and patient education

Four publications contain studies on information and patient education. One study had been conducted to gain insight into the several kinds of patient education materials on rare hereditary disorders that patient organisations

need. One article describes a school health education programme to provide information on sickle cell disease to children. Another investigation has been conducted to improve the information supply on rare diseases for both patients and their relatives. Both studies were carried out by an umbrella patient organisation. Finally, in one article the effects of professional and media warnings are discussed to promote or impede public awareness.

☞ Go to page 67 to find abstracts on information and patient education.

Two reports are written in Dutch, one journal article on general information and opinions and another (semi-)review article are written in English.

1.4 Outcome

1.4.1 Quality of care

The heading "quality of care" has been used in a broad sense. Publications included in this bibliography mainly address initiatives on quality improvement, such as the diagnostic process, as described in four articles. One article compares the formulation of diagnostic hypotheses by general internists and GPs. In a report, problems of patients suffering from rare diseases were investigated and recommendations given to improve the quality of care. One study compares health status (survival) of patients with rare diseases from 49 different health institutions. Furthermore, the GPs' knowledge on chronic diseases has been tested and, according to the authors, apparently this knowledge depends on practical experience. The less GPs had been confronted with a certain disease, e.g. a rare disease, the less knowledge they had about the disease. In only one study the concept of quality of care was measured as such.

☞ Go to page 70 to find abstracts on quality of care.

The 12 publications include one report, five research articles and six journal articles with general information or opinions. Ten publications are written in English and two in Dutch.

1.4.2 Quality of life

Quality of life is a term that is often used to describe one's well-being. In health care research, quality of life is usually a multi-dimensional concept with at least a somatic, psychological and a social aspect. Four studies were included in the bibliography in which overall quality of life was measured on

patients suffering from rare diseases, such as multiple sclerosis, celiac disease, Gaucher disease and scleroderma. One article discusses overall quality of life for children with Crohn disease.

☞ Go to page 79 to find abstracts on quality of life.

The five publications include three research articles, one report and one journal article with general information or opinions. Three publications are written in English, one in Dutch and one in Norwegian.

1.4.3 Psychosocial functioning and coping

Relatively many studies investigate psychosocial functioning and coping with a rare disease. The topic of psychosocial functioning can be considered as one of the dimensions of the concept "quality of life". The main goal of most studies was to find out how a patient gets along with a rare chronic progressive disorder (coping strategies), in order to gain insight into subjective experiences, adjustment and effects of the illness on social relationships. Also, the emotional and psychosocial effects on the parents of children with rare diseases or other family members were written down in different studies. Furthermore, two studies gave insight into the structure and determinants of illness representations or illness perceptions of patients suffering different rare diseases. One study focuses on methodological problems of the study, instead of the results of coping with a rare disease. Finally, two articles discuss sexual problems related to the rare disease.

☞ Go to page 83 to find abstracts on psychosocial functioning and coping.

The twenty-two publications include fourteen research articles, three review articles, one dissertation, one book (chapters) and three journal articles with general information or opinions. Twenty-one publications are written in English and one in German.

1.4.4 Social aspects

Like the topic of psychosocial functioning, social aspects can be considered as one of the dimensions of the concept "quality of life". This category was separated from psychosocial functioning or quality of life, because it mainly consisted of topics related to social relations and social problems of patients with rare diseases. In one study, the problems related to familial and professional reintegration of patients who had been treated for rare diseases are investigated. Similarly, one study assesses whether improvements in treatment had influenced the prospects of patients in the labour market. One

study explores the perceptions and experiences of lay care-giving. Furthermore, the family situation, mothers' health and financial assistance, such as social insurance were studied. Although studies on costs of specific rare diseases were excluded from the bibliography, one cost-study has been included. This study provides a model for the assessment of rare disease costs in general.

☞ Go to page 99 to find abstracts on social aspects.

From six publications, five are research articles and one is partly a review article. Five articles are written in English and one in French.

1.5 Arrangement of the bibliography

In the next chapter, the abstracts in the bibliography are divided into three main paragraphs: structure, process and outcome, and several subparagraphs, in which references to literature on different subjects can be found. In each subparagraph the references are listed in alphabetical order of the first author. Every article from a periodical to which the NIVEL library is not subscribed has a 'C'-number. Articles from periodicals to which the NIVEL library does have a subscription do not have a number. Books have 'B'-numbers and reports have 'R'-numbers. All publications are available and can be obtained from the NIVEL library.

1.6 Conclusion

This bibliography offers over one hundred journal articles, reports and documents on topics that are directly or indirectly related to daily life and health care for people with rare diseases. A rare disease used to be something extraordinary that could not be treated. Although probably most rare diseases can still not be treated or cured, the introduction of the Orphan Drug Act in the USA and Europe seems to have focussed the attention on rare diseases. The increasing number of documents in this bibliography that have been published in the last ten years reflects this growing awareness. It was, however, not easy to find relevant publications to include into this report:

An extensive search strategy has been performed and an enormous number of abstracts have been screened (N=3,241), according to eight exclusion criteria. Only a few publications, however, were considered relevant to be

included into the bibliography (N=118). It was striking to see that the number of research articles in the selected publications was rather low (N=41). Compared to the other topics, most research articles (N=14) that were found discussed psychosocial functioning and coping, but even these numbers are quite low. Besides research, 46 publications contained general information or opinions on matters such as health and Orphan Drugs policy. Moreover, sixteen reports (11 on research), three books (chapters), ten review articles or articles that partly review a disease, one governmental publication and one dissertation are included in the bibliography. Most publications are written in English, but also a relatively large number of Dutch documents have been found. This is probably due to easy accessibility to Dutch databases and libraries. Relatively more documents originate from European authors than non-Europeans. A relatively large number of publications are written by American and Dutch authors.

Although in recent years, people living with rare diseases are slowly gaining more attention, they still can be considered a forgotten group in social and scientific research. So far, research on the nature and scope of the consequences of living with a rare disease has been scarce. More research into the topics of this bibliography is necessary, to gain a more profound insight into quality of life, quality of care, coping with a rare disease, specific needs, or other topics, preferably from the patients' perspective. Besides medical treatment, which is often not available, people with rare diseases seem to have other health care needs as well. Hopefully in the near future, policy makers will become (even) more aware that the needs of people with rare diseases are urgent.

References

►EC: COMMISSION OF THE EUROPEAN UNION. Decision of the European Parliament and Council for a communal action programme 1999-2003 concerning rare diseases within the framework of public health. Luxemburg: Office for official publications of the European Union, 1997.
►NISPEN RMA van, RIJKEN PM & HEIJMANS MJWM. Leven met een zeldzame chronische aandoening. Ervaringen van patiënten in de zorg en het dagelijks leven. Utrecht, The Netherlands: NIVEL, 2003.
►ORPHANET: 2002. www.orpha.net
►ORPHANET: Final scientific report. Programme of community action on rare diseases. Orphanet, 2003.
►RGO: Raad voor Gezondheidsonderzoek. Advies Orphan Drugs (weesgeneesmiddelen). Rijswijk: Raad voor gezondheidsonderzoek, 1998.
►TULDER MW van, ASSENDELFT WJJ, KOES BW, BOUTER LM & The Editorial Board of the Cochrane Collaboration Back Review Group (1997). Method guidelines for systematic reviews in the Cochrane Collaboration back review group for spinal disorders. Spine, 22(20), 2323-2330.
► ROBINSON R. Managed Care: US evidence and lessons for Europe. Eurohealth 1998; 4 (2): 33-34.
► COLOMBO, F. Towards more choice in social protection? : individual choice of insurer in basic mandatory health insurance in Switzerland. Paris : OECD, 2001. (Labour market and social policy ; 53)
► EUROPEAN OBSERVATORY ON HEALTH CARE SYSTEMS. Health Care Systems in transition: Switzerland. WHO, 2000.

2 ABSTRACTS

2.1 Structure of health care

2.1.1 Health policy

Commission of the European Union
Decision of the European Parliament and Council for a communal action programme 1999-2003 concerning rare diseases within the framework of public health
Luxemburg: Office for official publications of the European Union 1997
[Voorstel voor een besluit van het Europees Parlement en de Raad tot vaststelling van een communautair actieprogramma 1999-2003 inzake zeldzame ziekten binnen het actiekader op het gebied van de volksgezondheid. Luxemburg: Bureau voor officiële publicaties der Europese Gemeenschappen 1997]
NIVEL: R 10290

In this report a proposal is given for an action programme concerning research and management of rare diseases. First, some important facts and problems concerning rare diseases are described, like a lack of expertise and of information for patients and their relatives. Next, an overview is given of proposed public health actions on an international, community level in the European Union. These actions include the creation of a European information system and networks on local, national and European levels, surveillance of rare diseases and the stimulation of patient organisations.

Language: English, Dutch
Publication Type: Report
Keywords: Health policy / European Union / International cooperation / Orphan drugs / Research

Garattini S
Orphan diseases and treatments including orphan drugs and some orphan indications
In: Fracchia GN & Haavisto KH (eds.): Perspectives in clinical trials. Paris; London: European
Commission, 1996, p. 117-124
NIVEL: C 8142

In this chapter the priorities for research regarding rare diseases are outlined. This is followed by an example of a clinical research centre, the Mario Negri Institute for Pharmacological Research in ltaly. This centre provides the following services: the Information Centre for Rare Diseases, the Clinical Research Centre and the European School for Rare Diseases. The Information Centre supplies patients and health care professionals with up-to-date information on the causes, prognosis and therapeutic possibilities for rare diseases. The Research Centre is a working clinic where patients with a rare disease are invited to spend a week, to study several patients with the same disease at the same time. The various projects represent an opportunity to improve on existing studies and to offer explanations about new findings to patients. The European School offers courses to train health care professionals.

Language: English
Publication Type: Book chapter
Keywords: Orphan drugs / Information services / Research / Italy

Groft SC
Rare diseases: identifying needs. Report of the National Commission on Orphan Diseases
American Pharmacy 1990; NS30(4):33-40
NIVEL: C 8137

This article reports of a survey, performed in 1985 by the National Commission on Orphan Diseases (NCOD) of the United States. Most patients with rare diseases want to be informed about their illness and available services. Physicians reported having difficulty obtaining information about rare diseases and their treatment to distribute to their patients. This paper also outlines the recommendations of the NCOD to improve delivery of information, research policy and development of orphan drugs.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Information need / Information supply / Orphan drugs / Research / Health policy

McCoy KS, Stokes D, Dubroff M, Hoffman A, Mischler NE, Erickson S
Managed care and the cystic fibrosis patient
Current Opinion in Pulmonary Medicine 1997; 3(6): 425-429
NIVEL: C 8138

This article addresses trends and policy in managed care for patients suffering cystic fibrosis. Physicians and consumers continue to have concerns about the potential negative impact of managed care on a relatively rare, specialised, and chronic illness such as cystic fibrosis if managed care concepts are applied without proper understanding of the disease. A great concern is that managed care in cystic fibrosis may cause reversal of trends in improved quality and length of survival. The increased length of survival places an increasing demand on the already stressed system of health care financing. Understanding the changing area of managed care is therefore of paramount importance to clinicians involved in cystic fibrosis care. The Cystic Fibrosis Foundation has presented symposia on managed care at each of the last three annual meetings, including the North American Cystic Fibrosis Meeting, October 1996. The following issues were addressed by speakers and panel discussants, with portions excerpted for this review: trends in managed care, measures and guidelines useful in managed care, Medicaid managed care and cystic fibrosis, and practical aspects of using pathways in caring for patients with cystic fibrosis.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Cystic fibrosis / Managed care / Health policy / United States / Quality of care

Taruscio D, Cerbo M.
Rare diseases: general principles, specific problems, and health interventions
[Malattie rare: concetti generali, problemi specifici ed interventi sanitari]
Annali dell'Instituto sLaperiore di sanita 1999;35(2): 237-244
NIVEL: C 8139

This article addresses some general concepts and specific problems regarding rare diseases as well as public health activities being undertaken in the USA, EU and ltaly. First, a short description is given of a few specific rare diseases, like Niemann-Pick disease, Behçet's disease or Prader-Willi syndrome. Next, typical problems regarding rare diseases are outlined, like lack of knowledge and late diagnosis. Finally, policy interventions in the EU, USA and, more elaborately, in ltaly are described. These interventions are aimed at national cooperation of expertise centres, patient education and accessibility of orphan drugs.

Language: Italian
Publication Type: Journal article, general information and opinions

Keywords: Health policy / Public health / European Union / United States / Italy / Niemann-Pick disease / Behçet's disease / Prader-Willi syndrome

Taruscio D, D'Agnolo G
Genetic diseases: recent scientific findings and health and ethical problems
[Malattie genetiche: recenti acquisizioni scientifiche e problemi sanitari ed etici]
Annali dell'Instituto superiore di sanita 1999;35(2):165-175
NIVEL: C 8140

This article deals with the ethical, legal and social problems concerning genetic screening for discovering rare genetic diseases, especially cystic fibrosis, Huntington's disease and muscular dystrophy of Duchenne. The Italian guidelines for genetic tests are described. In accordance with recommendations by international bodies (WHO, EU), the guidelines give emphasis to the appropriate use of both safe and efficacious tests, the performance in laboratories with high quality standards. A further crucial point is the relationship between the health system and individuals: autonomy of decision, psychological and social assistance, as well as adequate attention to ethical and privacy problems should be guaranteed.

Language: Italian
Publication Type: Journal article, general information and opinions
Keywords: Genetic diseases / Cystic fibrosis / Huntington disease / Muscular dystrophy, Duchenne / Genetic screening / Ethics, medical / Guidelines / Italy

2.1.2 Orphan drugs policy

Arno PS, Bonuck K, Davis M
Rare diseases, drug development, and AIDS: the impact of the Orphan Drug Act
The Milbank Quarterly 1995; 73(2): 231-252

In this article the history of the Orphan Drug Act is described, as well as its strengths and weaknesses. Although the act has achieved numerous successes, the high prices and extraordinary sales generated by some orphan drugs lead to a pivotal policy question: how can the act be used to meet the legislative goal of stimulating drug development for small patient populations without resulting in prices that make drugs inaccessible? This question is explored using the example of AIDS drugs, many of which received subsidies under the act, to illustrate central points. The authors

propose that the government grants a monopoly under 2 circumstances for drugs developed with public subsidies: 1) if it can be demonstrated that monopoly pricing and profits are needed to offset an extraordinary investment required to bring a new drug to market, and 2) if the potential market for the drug is so small that no company is willing or able to invest the requisite resources in research an development. Where these circumstances do not exist, the government should grant nonexclusive licensing agreements when it has been substantively involved in the development of a new drug.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Aids / Orphan drugs / Health policy / United States / Drug costs / Financing

Asbury CH
The Orphan Drug Act. The first 7 years
JAMA 1991; 265(7): 893-897

Objective: To compare changes in orphan drug production before and after the 1983 Orphan Drug Act which sought to increase market incentives (like market exclusivity, tax credits, and regulatory process clarifications) and decrease regulatory barriers for products used to treat rare ("orphan") diseases.
Methods: A comparison is made of the numbers of pre- and post-Act orphan products developed.
Results: While industry sponsored 34 marketed and 24 experimental orphan drugs in the 17 years prior to the Act, it has sponsored 39 of 42 marketed orphan products in the 7 years since the Act. An additional 301 experimental products have orphan designation. While 25 of 40 marketed orphan products reportedly had annual sales of less than $1 million, product sales for three conditions are more than $100 million annually. This prompted changes in the law, passed by Congress in 1990, but vetoed.
Conclusions: Overall, the law has been associated with an increase in orphan product development. The law's costs and benefits to companies, patients, and the public should be examined if future changes are proposed.

Language: English
Publication Type: Journal article, research
Keywords: Orphan drugs / Health policy / United States / Drug industry / Surveys

Borst-Eilers E
Coordination, priority setting and stimulation of research in the Netherlands concerning orphan drugs
[Coördinatie, prioritering en bevordering van onderzoek in Nederland op het terrein van de weesgeneesmiddelen voor zeldzame ziektes]
Den Haag, SDU, 2000
NIVEL: K 26800(16(80

In this letter the Dutch minister of public health describes shortly the policy of the European Union regarding research and development of orphan drugs. Next recommendations concerning coordination and priority setting in the field of orphan drugs are outlined. These recommendations are not only nationally oriented, but also on a European level. A national organisation is needed for collecting information, coordinating and stimulating research. This organisation should be funded by the Dutch government.

Language: Dutch
Publication Type: Official document of the Dutch government
Keywords: Orphan drugs / Health policy / Research / The Netherlands / European Union

Brandissou S, Yagoubi N, Hasselot N
Orphan drugs: a problem of public health and an economic stake
[Les médicaments orphelins: problème de santé publique et enjeux économiquesj
Therapie 1996; 51: 647-653
NIVEL: C 8141

In this article an outline is given of the regulation concerning orphan drugs (OD). The United States initiated work in order to confer status in OD leading to the Orphan Drug Act in 1983. It defined the conditions for attribution of the OD status and also made attractive proposals to pharmaceutic industries in order to improve their development. More recently, in 1993, Japan took similar decisions, while the European Union and France are also on the way, as recent meetings of European ministers of health show. In contrast, developing countries are still excluded from medical research as very few tropical diseases have treatment. Legislation for OD, first planned to make up for the high costs of research and development, proved its efficiency. But ODs must move to a new status as some are now becoming the object of important economic stakes.

Language: French
Publication Type: Journal article, general information and opinions

Keywords: Orphan drugs / Legislation / United States / France / European Union / Economics

Eijkel J van
Second report on orphan drugs and the abstract and conclusions from the first report on orphan drugs
[Vervolg-rapport Weesgeneesmidddelen en de samenvatting en conclusies uit de 1ste rapportage Weesgeneesmiddelen]
Amstelveen: College voor Zorgverzekeringen, 2003
NIVEL: R 9400(146

This report is a sequel to the report 'Admittance of orphan drugs to the social insurance package' by Zwaap (see below). In this second report recommendations are given on an evaluation procedure for including orphan drugs in the social insurance package in the Netherlands. The abolishment of the requirement of testing a new drug by pharmaco-economic research is considered. It is concluded that the criterion of pharmaco-economic research does not have to be met in case there are not yet sufficient treatment methods for the disease in question. Indications have to be less frequent than 1 to 150000 inhabitants to meet the criterion of rareness of the disease, and the drugs are not to be registered in the Netherlands.

Language: Dutch
Publication Type: Report
Keywords: Orphan drugs / Social insurance / Health policy / Reimbursement / Monitoring / The Netherlands

Lang J, Wood SC
Development of orphan vaccines: an industry perspective
Emerging Infectious Diseases 1999; 5(6): 749-756
NIVEL: C 8143

This article addresses the problem that the development of vaccines against rare emerging infectious diseases is hampered by many disincentives. In the face of growing in-house expenditures associated with research and development projects in a complex legal and regulatory environment, most pharmaceutical companies prioritise their projects and streamline their product portfolio. Nevertheless, for humanitarian reasons, there is a need to develop niche vaccines for rare diseases not preventable or curable by other means. The U.S. Orphan Drug Act of 1983 and a similar proposal from the European Commission (currently under legislative approval) provide financial and practical incentives for the research and development of drugs

to treat rare diseases. In addition, updated epidemiologic information from experts in the field of emerging diseases; increased disease awareness among health professionals, patients, and the general public; a list of priority vaccines; emergence of a dedicated organisation with strong leadership; and the long-term pharmaco-economic viability of orphan products will be key factors in overcoming the complexity of orphan status and the limited need tor vaccine.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Orphan drugs / Vaccines / Health policy / United States / European Union

Linthorst GE, Hollak CEM
European ordinance on orphan drugs: changes and threats
[Europese verordening inzake weesgeneesmiddelen: kansen en bedreigingen]
Nederlands Tijdschrift voor Geneeskunde 2003; 147(4): 143-145

In this article an explanation is given of the European ordinance on orphan drugs. Next, the problems of reimbursement of orphan drugs in the Netherlands are outlined. The Orphan Drug Act has been officially implemented in all countries of the European Union since the year 2000. The Act aims to promote the development of treatments for rare diseases. Successful therapies for such diseases are granted market-monopoly for 10 years, during which no other drug for the treatment is allowed. There are problems concerning approval and reimbursement. For instance, drugs already in use can also be registered as orphan drugs for a specific indication. In addition, European approval for market authorisation of new drugs does not automatically mean that the costs of the drugs are reimbursed within the Netherlands, even if that is the case in other EU countries. However, a faster procedure with respect to the reimbursement of drug costs in the Netherlands may not lead to the acceptance of new drugs before additional research trials being carried out. Therefore, a national organisation, funded by the government, is installed to stimulate the drug development and to enhance information supply for patients and relatives.

Language: Dutch
Document Type: Journal article, general information and opinions
Keywords: Orphan drugs / Health policy / The Netherlands / European Union / Insurance / Reimbursement

Meijer I, Vos R, de Jong-van den Berg LTW, Meijer D
Orphan drugs
In: Timmerman H, Berg Jeths A van den. Geneesmiddelen nu en in de toekomst: Volksgezondheid Toekomst Verkenning: achtergrondstudie. Houten: Bohn Stafleu van Loghum, 2001
NIVEL B 5000 (book)

In this chapter a short description is given of the prevalence and incidence of rare diseases, followed by an outline of the legislation of orphan drugs in the European Union and the United States. Next, the viewpoints of patient organisations, pharmaceutical industry, universities and government on the development of orphan drugs are described. The chapter ends with the research effects regarding orphan drugs and possibilities for the future.

Language: Dutch
Publication Type: Reports
Keywords: Orphan drugs / Legislation / Health policy / European Union / United States / Research

Minghetti PM, Giudici EM, Montanari L
A proposal to improve the supply of orphan drugs
Pharmacological Research 2000; 42(1): 33-37
NIVEL: C 8144

In this article, the regulations concerning the supply of orphan drugs, enforced in the USA, Japan, and the European Union are discussed, as well as the use of possible incentives to the industry. In the United States, a specific legislation on orphan drugs dates back to 1983; in the European Union, regulations are currently being studied. The industrial production of orphan drugs might be improved following the adoption of adequate incentives. New regulations should also take care of neglected doses, and stimulate the preparation of galenic products. Galenic products can represent an effective alternative to the industrial product; they can guarantee immediate availability of dosages and pharmaceutical forms which are not available in the industrial market.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Orphan drugs / Health policy / United States / European Union / Japan

Raad voor Gezondheidsonderzoek
Recommendations concerning Orphan Drugs
[Advies Orphan Drugs (weesgeneesmiddelen)]
Rijswijk: Raad voor gezondheidsonderzoek, 1998
NIVEL: R 9406

This report, by the Dutch Advisory Health Research Council (RGO), offers recommendations on the coordination, prioritisation and advancement of research in the field of orphan drugs in the Netherlands. A study was made of the expertise, research facilities and bottlenecks in Dutch research concerning orphan drugs. The Council recommends the establishment of an integrated policy with a national coordinating body in which scientific investigators, patient organisations, pharmaceutical industry and government participate. The integration concerns research, development and finance, both national and European, with the aim to enhance cooperation between the just mentioned parties. A study is also performed to health care services needs in order to improve the balance between demand and supply. Moreover, 5 orphan diseases and their appropriate drug treatment are described.

Language: Dutch
Publication Type: Report
Keywords: Orphan drugs / Health policy / The Netherlands / European Union / Financing / Legislation / Health services needs

Roeleveld RJ, Postmus PE, Boonstra A
An expensive drug: ambulant treatment of pulmonary hypertension with epoprostenol
[Een duur medicament: pulmonale hypertensie ambulant behandelen met epoprostenol]
Medisch Contact 2002; 57(16): 634-637

This article addresses the problem of the lack of legislation concerning the financing of expensive drug treatment of rare diseases. An example is described of the development of a special financing system for the drugs which are used in treating primary pulmonary hypertension, a rare disease with a very poor prognosis which can be improved with these expensive drugs.

Language: Dutch
Publication Type: Journal article, general information and opinions
Keywords: Primary pulmonary hypertension / Orphan drugs / Financing / Health policy / The Netherlands

Seeverens HJJ
Medicines for rare diseases (‘orphan drugs')
[Geneesmiddelen voor zeldzame ziekten (‘orphan drugs')]
Geneesmiddelen bulletin 2001; 35(5): 49-53

This article addresses the problem of a shortage of drugs for effective treatment of rare diseases. A description is given of the policy concerning orphan drugs in both the United States and the European Union. Finally the Dutch policy is shortly described.

Language: Dutch
Publication Type: Journal article, general information and opinions
Keywords: Orphan drugs / Health policy / European Union / United States / The Netherlands / Registries

Sinnema M
The adoption of orphan drugs: the example of lyposomal storage diseases
[De adoptie van weesgeneesmiddelen: lyposomale stapelingsziekten als voorbeeld]
Utrecht: Wetenschapswinkel Geneesmiddelen, Faculteit Farmaceutische Wetenschappen, Universiteit Utrecht, 2001
NIVEL: R 10168

This report addresses the European and Dutch policy concerning the development of orphan drugs. This policy includes measures like incentives, financial aids and market exclusivity to stimulate the pharmaceutical industry. The issue in illustrated with an example of lyposomal storage diseases, a group of hereditary disorders, including Fabry disease.

Language: Dutch
Publication Type: Report
Keywords: Lyposomal storage diseases / Fabry disease / Orphan drugs / Health policy / Pharmaceutical industry / European Union / The Netherlands

Sinnema M, Geffen ECG van, Weely S van, Leufkens HGM
Another issue concerning rare diseases: the development of orphan drugs
[Een andere problematiek rond zeldzame aandoeningen; de ontwikkeling van weesgeneesmiddelen]
Pharmaceutisch Weekblad 2002; 137(32): 1116-1120

This article gives an overview of health policy measures concerning the development of orphan drugs in the Netherlands and the European Union. Problems of the pharmaceutical industry are described. Different parties in health care, including pharmacists and representatives of patient organisations, health insurance companies, the Dutch government and the pharmaceutical industry, cooperate in stimulating research, diffusion of knowledge and the development of special regulations regarding orphan drugs.

Language: Dutch
Publication Type: Journal article, general information and opinions
Keywords: Orphan drugs / Health policy / Drug industry / Pharmacists / The Netherlands / European Union

Thamer M, Brennan N, Semansky R
A cross-national comparison of orphan drug policies: implications for the U.S. Orphan Drug Act

Journal of Health Politics, Policy and Law 1998; 23(2) :265-290
Objective: To compare public policies of six countries - Canada, France, Japan, Sweden, the United Kingdom, and the United States - affecting the development and marketing of pharmaceuticals for rare diseases (i.e., orphan drugs).
Methods: Information was obtained from a variety of published and unpublished sources, including interviews with public policy and pharmaceutical experts in each country.
Results; This article presents different approaches to encouraging the development of orphan drugs while ensuring access by regulating their prices. Additionally, the article describes access to orphan drugs as promoted by special coverage for population subgroups, disease categories, and/or specific drugs. Not all efforts to increase access to orphan pharmaceuticals have been the result of government action, as illustrated by the proliferation of for-profit organisations that specialise in orphan drugs.
Conclusions: The many policy options from other countries identified in this study are especially relevant, given increasing calls for reform of the U.S. Orphan Drug Act.

Language: English
Publication Type: Journal article, research
Keywords: Orphan drugs / Health policy / Legislation / Pharmaceutical industry / Comparative study / Financing / Canada / France / Japan / Sweden / United Kingdom / United States

Zwaap J
Admitting orphan drugs into the social insurance package
[Toelating weesgeneesmiddelen tot het sociale verzekeringspakket]
Amstelveen: College voor Zorgverzekeringen, 2002
NIVEL: R 10249

In this report recommendations are given for the evaluation procedure for including orphan drugs in the social insurance package in the Netherlands. There is a difference between orphan drugs for which their therapeutic value and suitability are sufficiently known and orphan drugs for which this is not the case. The first group of orphan drugs can be evaluated by means of standard procedures. The other group of drugs has to be excluded from the social insurance package, basically. But these drugs have to be available on a special financial basis. This report also addresses the way orphan drugs can be monitored.

Language: Dutch
Publication Type: Report
Keywords: Orphan drugs / Social insurance / Health policy / Reimbursement / Monitoring / The Netherlands

2.1.3 Databases and surveillance systems

Calhoun RD
Practice guidelines for patients with rare cancers
Cancer Practice 1998; 6(4): 247-250
NIVEL: C 8175

This article offers guidelines for clinicians to offer disease-oriented information to patients suffering rare cancers. Internet resources are mentioned and strategies are outlined to build support networks.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Cancer / Information services / Patient education / Self-help groups

Elliott EJ, Chant KG
Rare disease surveillance
Journal of Paediatric Child Health 1994; 30(6): 463-465
NIVEL: C 8147

In this article some methods for the surveillance of rare diseases in children are described. Rare diseases in children account for disproportionate morbidity and mortality and are particularly demanding of both families and health resources. Surveillance may provide data on their epidemiology, aetiology, management and outcome and on the support requirements of affected children. Existing methods for rare disease surveillance include mandatory and voluntary notification schemes, which may be active or passive, hospital discharge databases and death certification data. The recent establishment of the Australian Paediatric Surveillance Unit has facilitated active, prospective, national case ascertainment by voluntary notification of selected rare conditions. Information obtained should enable estimation of incidence rates, evaluation of prevention and management strategies, extend data collected by existing methods and help estimate future health needs.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Children / Monitoring / Epidemiology / Australia

Hira Sing RA, Rodrigues Pereira R
The Dutch Paediatric Surveillance System: a quality-focussed instrument for prevention and research
[Het Nederlands Signaleringscentrum Kindergeneeskunde: een kwaliteits-instrument voor preventie en onderzoek] Nederlands Tijdschrift voor Geneeskunde 2002; 146 (50): 2409-2414

In this article the aims, activities and results of the Dutch Paediatric Surveillance System are described. This system was initiated by the Dutch Paediatric Society and is housed within the TNO Prevention and Health office. lts purpose is (a) to gain insight on a population level into the prevalence of rare and new diseases in youths (0-18 year), and (b) to promote scientific research addressing the background, nature and prognosis, as well as the treatment and prevention, of these diseases. A monthly report card is sent to all clinically active paediatricians with maximally 10 diseases under surveillance. The response rate is approximately 90%. The information from the registration can lead to primary or secondary prevention (prevention of the disease or of its complications, respectively). The system provides information for the amelioration of quality of care, for health management and, at the same time, acts as an instrument for scientific research.

Language: Dutch
Publication Type: Journal article, general information and opinions

Keywords: Children / Epidemiology / Monitoring / Research / Registries

Kindler H, Fliedner TM, Densow D
Internet access to a medical case repository for teaching and analysis
Journal of the American Medical Informatics Association 1997; volume unknown: 543-547
NIVEL: C 8151

This article describes the development and implementation of an information system for standardised recording of acute radiation syndrome case histories, which has been established in the framework of the World Health Organisation Radiation Medical Emergency Preparedness and Assistance Network Centres. Such an information system was considered necessary because the acute radiation syndrome is a rare disease; teaching its treatment and the achievement of scientific progress require a sufficient number of accessible case histories. The information system has been realised in client/server architecture. The centres are able to access the database by easy to use graphical user interfaces via the Internet. Focussing on optimisation of network traffic and flexible information retrieval the hypertext transfer protocol has not been applied.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Acute radiation syndrome / Information systems / Internet / Computer communication networks / Databases

Miravitlles M, Vidal R, Barros-Tizon JC, Bustamante A, Espana PP, Casas F, Martinez MT, Escudero C,
Jardi R
Usefulness of a national registry of alpha-l-antitrypsin deficiency: the Spanish experience
Respiratory Medicine 1998; 92(10): 1181-1187
NIVEL: C 8152

Objective: To describe the first 5 years of experience of a registry of severe alpha-l-antitrypsin (AAT) deficiency, phenotype Pi ZZ, a rare condition with an estimated prevalence of 1/4500 individuals in Spain. Given this low prevalence, it seemed useful to accumulate all the information derived from the care of these patients. In this context, the Spanish Registry of patients with AAT deficiency was founded in 1993; its main objectives were to establish guidelines adapted to our country for the treatment and

management of AAT-deficient patients, offer expert support to physicians all over the country treating these patients, and provide technical support on the determination of Pi phenotyping and genotyping of individuals suspected of being AAT-deficient.
Methods: From 1993 to January 1998 the number of enrollees increased from 48 to 223, of which 216 were Pi ZZ. Seventy-three per cent were male and only 31.5% were never smokers, mean age was 46 years (SD=13) and mean FEV1 53% predicted (SD=31). 83% were index cases who, compared with non-index cases, were older (49 +/- 11 vs. 35 +/- 13 years, $p<0.001$), more likely to have a smoking history (85% vs. 47%, $p<0.01$) and displayed more severe impairment in pulmonary function (FEV1% = 40% +/- 19% vs. 96% +/- 23%, $p<0.001$). Augmentation therapy was administered to 129 patients (58%).
Results: Treated patients had more severe impairment in pulmonary function than the untreated (FEV1% = 40% +/- 21% vs. 72% +/- 32%, $p<0.001$) and were more likely to be index cases (81% vs. 43%, $p<0.001$). Characteristics of the patients included are similar to those described for other Registries.
Conclusions: The Registry has extended knowledge of the disease throughout the country and has established local guidelines for treatment and follow-up. It may be a valid database for future cooperation in international initiatives.

Language: English
Publication Type: Journal article, research
Keywords: Alpha-I-antitrypsin deficiency / Registries / Databases / Guidelines / Spain / Evaluation study

Morris A, Ridley GF, Elliott EJ
Australian Paediatric Surveillance Unit: progress report
Journal of Paediatric Child Health 2002; 38(1): 8-15
NIVEL: C 8153

This article describes the role and progress of the Australian Paediatric Surveillance Unit (APSU), which, through active surveillance, collects information on the epidemiology of rare or uncommon childhood conditions. This research resource allows paediatricians to collaborate at State, national and international levels. The APSU sends a monthly report card to all (currently 971) paediatricians in Australia, who in turn indicate whether or not they have seen a patient in the last month with any of the conditions listed (98% response rate in 1999). Study investigators, notified by the APSU of positive case reports, obtain demographic and clinical data on the patient from the reporting doctor by postal questionnaire (90% response rate

in 1999). Since 1993, the APSU has monitored 27 conditions, including conditions that are vaccine-preventable, otherwise infectious, genetic, congenital and non-communicable. Information collected is disseminated to paediatricians and other health professionals via an annual report, newsletters and publications and is made available to the International Network of Paediatric Surveillance Units. Information provided by the APSU has raised awareness among paediatricians of rare and uncommon childhood conditions, and has been used by health authorities for planning of prevention, intervention strategies and allocation of health resources.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Children / Research Centres / Monitoring / Australia

Thoene JG, Coker NP (eds)
Physicians' guide to rare diseases
Montvale NJ: Dowden Publishing Company, 1995 (2nd edition)
Available in Groningen University Library

The aim of this reference work is to assist physicians who encounter rare diseases infrequently by providing ready access to signs and symptoms on rare diseases in order to help in differential diagnosis and the availability of therapy. This work is an adaptation of the rare diseases database entries for close to 700 rare diseases. Each major section opens with an overview article, followed by short disease descriptions, which include symptoms, etiology, treatment and information resources.

Language: English
Publication Type: Reference work
Keywords: Reference books

2.1.4 Research methodology

Capella D, Laporte JR, Vidal X, Wiholm BE, Begaud B, Langman MJ, Rawlings M
European network for the case-population surveillance of rare diseases (Euronet): a prospective feasibility study
European Journal of Clinical Pharmacology 1998; 53(5): 299-302
NIVEL: C 8202

Objective: To assess the feasibility of developing a case-population surveillance network (Euronet) for monitoring commonly iatrogenic rare

conditions, with regard to previous use of drugs and new active substances. This network aims to estimate the risk of certain rare conditions by comparing drug use amongst non-selective cases with overall drug use in the general population.
Methods: The method is based on three provisos: (1) all incident cases (irrespective of suspected aetiology) should be ascertained and studied; (2) a full drug history should be obtained from cases by direct interview; and (3) drug-use data for the products of interest should be available for this population from which cases are chosen. The feasibility of this problem oriented approach for the identification of new signals of adverse drug reactions and for risk estimation will be tested in relation to agranulocytosis, Stevens-Johnson syndrome and toxic epidermal necrolysis in four defined areas in Europe, totalling 19 million inhabitants, with these latest two outcomes being studied in only three regions. The design, methods and main limitations of this case-population surveillance approach are described.

Language: English
Publication Type: Journal article, research
Keywords: Agranulocytosis / Stevens-Johnson syndrome / Toxic epidermal necrolysis / Adverse drugs reactions / Registries / Population surveillance

De Groen PC, Barry JA, Schaller WJ
Applying World Wide Web technology to the study of patients with rare diseases
Annals of Internal Medicine 1998; 129(2): 107-113
NIVEL: C 8203

This article describes the development and implementation of a computerised disease tracing system coupled with a database accessible on the Web to study cholangiocarcinoma, a rare cancer of the bile ducts. The development of such a system was considered necessary because the World Wide Web allows efficient dissemination and exchange of information in order to conduct randomised, controlled trials of sporadic diseases. Unfortunately there is a lack of software for the identification of patients with a rare disease and subsequent data entry and analysis in a secure Web database. The tracing system scans computerised information systems on a daily basis and forwards demographic information on patients with bile duct abnormalities to an electronic mailbox. lf informed consent is given, the patient's demographic and preexisting medical information available in medical database servers are electronically forwarded to a UNIX research database. Information from further patient-physician interactions and procedures is also entered into this database. The database is equipped with a

Web user interface that allows data entry from various platforms (PC-compatible, Macintosh, and UNIX workstations) anywhere inside or outside our institution. To ensure patient confidentiality and data security, the database includes all security measures required for electronic medical records. The combination of a Web-based disease tracing system and a database has broad applications, particularly for the integration of clinical research within clinical practice and for the coordination of multicentre trials.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Bile duct cancer / Cholangiocarcinoma / Internet / Software / Databases / Medical records systems

Grufferman S, Delzell E, Delong ER
An approach to conducting epidemiologic research within cooperative clinical trials groups
Journal of Clinical Oncology 1984; 2(6): 670-675
NIVEL: C 8204

Objective: Cooperative clinical trials groups offer exciting opportunities for conducting epidemiologic research for several reasons: they facilitate accrual of sufficient numbers of subjects in a short period of time, even for studies of rare diseases; they provide uniform pathologic review and uniform collection of subjects' entry data; and they provide a more representative sample of cases than a single-institution study. Despite these advantages, few epidemiologic studies of etiologic factors have been done through these groups because methods for selecting appropriate control subjects and for obtaining information from geographically scattered subjects have not been available.
Methods: An approach that can serve as a model for this type of research has been developed. A collaborative case-control study of childhood rhabdomyosarcoma (RMS) with the Intergroup Rhabdomyosarcoma Study (IRS) was recently begun. The study, which is independently funded, evaluates the role of environmental factors in the etiology of RMS. Parents of subjects were interviewed by telephone and control subjects were selected from the same communities as patients by random digit dialing. Interview data are supplemented by information from birth certificates and, for patients, by IRS data.
Results and conclusions: This new methodology permits a large study of a rare tumor in a relatively short period of time.

Language: English
Publication Type: Journal article, research
Keywords: Rhabdomyosarcoma / Children / Clinical trials / Epidemiology / Research design / Interview study

Haffner ME
Designing clinical trials to study rare disease treatment
Drug Information Journal 1998; 32: 957-960
NIVEL: C 8205

This paper provides an overview of the design of clinical trials for rare diseases and the impact of the Orphan Drug Act on the successful study of diseases found in small, widely dispersed patient populations. Randomised, placebo-control trials may not always be an option for studying orphan drugs. Designs which may work better are discussed: open protocol, open label, historical control, crossover trials, withdrawal design trials, or trials with surrogate endpoints. The benefits of studying orphan drugs and the procedure for approval of trial design by the Food and Drug Administration are outlined.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Orphan drugs / Clinical trials / Research design

Honkanen VE, Siegel AF, Szalai JP, Berger V, Feldman BM, Siegel JN
A three-stage clinical trial design for rare disorders
Statistics in Medicine 2001; 20(20): 3009-3021
NIVEL: C 8206

Objective: To design a clinical trial method with improved statistical power compared to the traditional randomised trial for use in clinical trials of rare diseases.
Methods: A three-stage clinical trial design was used, consisting of an initial randomised placebo-controlled stage, a randomised withdrawal stage for subjects who responded, and a third randomised stage for placebo non-responders who subsequently respond to treatment. This design was compared with the traditional randomised trial design. Test level and power were assessed by computer-intensive exact calculations.
Results: The three-stage clinical trial design was found to be consistently superior to the traditional randomised trial design in all cases examined, with sample sizes typically reduced by 20% to 30% while maintaining

comparable power. When a treatment clearly superior to placebo was considered, our design reached a power of 75% with a sample of 21 patients compared with the 52 needed to attain this power when only a randomised controlled trial was used.
Conclusions: In situations where patient numbers are limited, a three-stage clinical trial design may be a more powerful design than the traditional randomised trial for detecting clinical benefits.

Language: English
Publication Type: Journal article, research
Keywords: Research designs / Clinical trials / Three-stage design

Kozinetz CA, Skender ML, MacNaughton NL, del Junco DJ, Almes MJ, Schultz RJ, Glaze DG, Percy AK
Population-based registries using multi disciplinary reporters: a method for the study of pediatric neurologic disorders
Journal of Clinical Epidemiology 1995; 48(8): 1069-1076

Objective: To present standard methodologies for establishing a population-based registry and evaluating the completeness of a registry's case ascertainment for rare, newly, or ill-defined pediatric neurologic disorders.
Methods: The Texas Rett Syndrome Registry (TRSR) is used as a model. Physician reporters were identified and registered and telephoned to request their participation as reporter for the TRSR.
Results: The combination of health care and education resources has identified approximately 89-100% of the Rett syndrome cases in Texas. Cases reported by non-physician sources, although older on average (10.7 versus 7.7 years of age), did not differ by other demographic characteristics from those reported by physicians. Non-physician health and education professionals participated with the TRSR at a significantly higher rate than physicians, 89 and 37% ($p<0.05$), respectively. Capture-recapture techniques, both two-sample and log-linear modelling, were used to quantitatively evaluate case ascertainment.
Conclusions: Standardised national and international population-based registries could be the basis of an initiative to identify the etiology and perhaps preventive measures for pediatric neurologic disorders.

Language: English
Publication Type: Journal article, research
Keywords: Rett syndrome / Nervous system diseases / Registries / Epidemiology / Linear models / Statistical models / Population surveillance

Lilford RJ, Thornton JG, Braunholtz D
Clinical trials and rare diseases: a way out of a conundrum
BMJ 1995; 311(7020): 1621-1625

In this article the problem is addressed of randomised clinical trials when the treatment of a rare disease has to be evaluated. Bayesian methods utilise all available data to calculate probabilities that may be extrapolated directly to clinical practice. The Bayesian approach provides probabilities of treatment effects that apply directly to the next patient who is similar to those treated in any completed or ongoing trial. These probabilities are calculated on the basis of the observed data and a prior distribution of probabilities. A trial can be analysed as data accumulate. This approach is illustrated with an example.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Clinical trials / Bayesian statistics

Rzany B, Mockenhaupt M, Baur S, Schroder W, Stocker U, Mueller J, Hollander N, Bruppacher R, Schopf E
Epidemiology of erythema exsudativum multiforme majus, Stevens-Johnson syndrome, and toxic epidermal necrolysis in Germany (1990-1992): structure and results of a population-based registry
Journal of Clinical Epidemiology 1996; 49(7):769-773

Objective: This article reports on the severe skin reactions erythema exsudativum multiforme majus (EEM with mucosal involvement, EEMM), Stevens-Johnson syndrome (SJS), and toxic epidermal necrolysis (TEN). These skin reactions are difficult to study as they are very rare diseases with an incidence of about two cases per 1 million inhabitants per year. The authors report on the structure of a registry with the aim of ascertaining all hospitalised cases of EEMM, SJS, and TEN in western Germany and Berlin.
Methods: The registry is structured as an intensive reporting system, regularly contacting more than 1500 departments including 100% of the burn units (N=34), departments of pediatrics (N=241), departments of dermatology (N=106), and 100% of all internal medicine departments in hospitals with intensive care facilities or with more than 200 beds (N=1161).
Results: With a coverage rate up to 95% based on the number of responding departments between April 1, 1990 and December 31, 1992, from a total of 767 reported cases 353 patients with EEMM, SJS, and TEN were finally included in the registry. Most of these patients were directly reported to the registry; only 2.54% (9 of 353) were primarily registered by the German spontaneous reporting systems. Assuming an average population of 64.5

million for western Germany and Berlin an incidence up to 1.89 per 1 million inhabitants per year could be calculated for SJS and TEN.
Conclusions: These data demonstrate a high acceptance of the registry and allow a variety of analyses. By increasing the number of well-documented cases reported to health authorities and by providing risk estimates for certain drugs, the registry performs public health functions and improves drug safety in Germany.

Language: English
Publication Type: Journal article, research
Keywords: Erythema multiforme / Stevens-Johnson syndrome / Toxic epidermal necrolysis / Adverse drugs reactions / Registries / Epidemiology / Germany

Wilcken B
Rare diseases and the assessment of intervention: what sorts of clinical trials can we use?
Journal of Inherited Metabolic Disease 2001; 24(2): 291-298
NIVEL: C 8207

This article addresses the problem that, for rare diseases, there are great difficulties in obtaining adequate evidence for interventions or for the benefits of early diagnosis, while randomised controlled trials are the standard way. The disorders are not only very rare but also have variable expression, may have very long courses, and have incompletely known late effects; and surrogate end-points often have to be used. Randomised controlled trials are usually impossible because of inadequate power, and because there are preconceived notions of the effects of treatments already in use. The author suggests the adoption of the best possible design for observational trials, formation of a central registry of such trials, and a greater general appreciation of the problems that rare diseases pose will help in obtaining the best possible evidence for the effects of interventions.

Language: English
Publication Type: Review
Keywords: Research design / Clinical trials

Yeaworth RC
Use of the Internet in survey research
Journal of Professional Nursing 2001; 17(4): 187-193
NIVEL: C 8145

Objective: To get insight into the potential for use of the Internet for research into diseases that are too rare to find sufficient subjects to gather data on patient and family experiences, helpful interventions, and other care-related information. In this case, a survey is conducted about Pick's Disease.
Methods: An author-constructed instrument posted on a university server was used to collect information on both the affected persons and the family.
Results: Although the sample cannot be scientific and controlled, there was a 68.6% response rate; 59 questionnaires were returned, of which 56 could be used. The fact that specialists and imaging technology had been used in making the diagnoses, and that, with one exception, the responses were comparable to information in other reports, increased confidence in the validity of the sample and the responses. One of the biggest needs of caregivers and family members was for more information and support.
Conclusions: This survey indicates that such research can provide information about rare diseases so that nurses can be more helpful to patients and caregivers. Further exploration of research uses of the Internet could not only help to get information on rare diseases, but perhaps offer unique research opportunities to students who are using distance-education technologies.

Language: English
Publication Type: Journal article, research
Keywords: Pick's disease / Internet / Diagnosis / Psycho social problems / Caregivers / Information need / Quality of care / Survey / Questionnaire study

2.2 Health care processes

2.2.1 Continuity and coordination of care

Berntorp E, Lethagen S, Ljung R, Johnsson H, Petrini P, Schulman S, Tengborn L, Rehnby B, Sarman E, Asberg B
Centralised care is the basis of care programmes for hemophilia patients
[Centraliserad vård grundlåggande i vårdprogram för blödarsjuka]
Lakartidningen 1999; 96(15): 1849-1852
NIVEL: C 8156

This article describes a care programme which has been designed to guarantee Swedish haemophiliacs comparable and optimal care. The programme has been drawn up by representatives of the three haemophilia centres in Sweden (at University Hospital, Malmo, Sahlgrenska University Hospital, Gothenburg, and Karolinska Hospital, Stockholm) in cooperation

with the World Federation of National Haemophilia Organisations. To ensure optimal individual application of the programme, individualised management strategies and patient information leaflets have been prepared. Attention is also paid to psycho social aspects.

Language: Swedish
Publication Type: Journal article, general information and opinions
Keywords: Haemophilia / Centralised hospital services / Patient education

Boyd JR, MacMillan LJ
Multiple sclerosis in childhood: understanding and caring for children with an "adult" disease
Axon 2000; 22(2): 15-21
NIVEL: C 8157

In this article multiple sclerosis (MS) in childhood is reviewed. The disease is rare in childhood but may occur more frequently than originally believed. In light of the complex nature of MS, the expanding availability of new MS treatments, and the developmental needs of paediatric patients, multi disciplinary care of paediatric patients with MS is a necessity. This review aims to increase the awareness of neuroscience nurses about MS in children and adolescents, and to expand the knowledge of paediatric nurses concerning this "adult" disease. The authors' experience in developing a multi disciplinary pediatric MS clinic that addresses both health and developmental needs of children with MS is presented and discussed. The nurse's role in the care of these children and their families through assessment, support, education, advocacy, referral and coordination of care is emphasised.

Language: English
Publication Type: Journal article, review
Keywords: Multiple sclerosis / Children / Adolescents / Multi disciplinary care / Coordination of care / Nursing / Centralised hospital services

Brummelen SE van
Health care for patients with rare diseases
[Zorg voor patiënten met zeldzame ziektebeelden]
Utrecht: Vereniging Academische Ziekenhuizen, 2000
NIVEL: R 10293

This report, aimed at advising academic hospitals in improving quality of care, describes the problems in the care of patients suffering rare diseases, from the patient perspective. Problems after diagnosing the illness, such as too many referrals, failure to recognise complications, lack of expertise and knowledge on patient organisations, are emphasised. Concentrating and cooperating health care services maintain a possible solution.

Language: Dutch
Publication Type: Report
Keywords: Continuity of care / Centralised hospital services / Patient education / Patient satisfaction / Academic hospitals

Chantler C
Paediatric nephrology on the threshold of European integration
Pediatric Nephrology 1991; 5(4): 428-432
NIVEL: C 8158

This article addresses the need for international cooperation in the care of children with kidney disease. Since the foundation of the European Society for Paediatric Nephrology (ESPN) 25 years ago, the progress of paediatric nephrology in Europe has been considerable, but now there are a number of problems. The care of the child with kidney disease is often expensive and therefore, more needs to be done to examine the cost effectiveness of the management of the child with kidney disease. End-stage renal failure is rare in childhood and likely to become even less common in the future. International co-operation can also foster clinical research to determine the effectiveness of treatment through the institution of controlled trials and outcome studies. Particular problems are posed by the need to integrate the countries of Eastern Europe that have changed from command to market economies over the last year. In many instances there is no shortage of doctors, nurses and hospital beds but there is a need to change administrative and academic structures and to introduce appropriate technology. It is suggested that this may be assisted by twinning units. The integration of Europe requires that the role of the paediatric nephrologist in different countries needs to be examined, and appropriate training to fulfill these responsibilities needs to be agreed. Different countries obviously have different ways of organising and providing services for children with kidney disease. Kidney failure is rare in childhood and there are economic and academic advantages from close collaboration, both with adult nephrology services and with other paediatric specialities. Where it is intended to integrate children's hospitals into large multi disciplinary university hospitals for economic reasons, it is nonetheless necessary to make sure that

which has been set up at the Centre for Disease Control, in Stockholm, and University Hospital, Linkoping, in collaboration with the Board of Health and Welfare, has introduced a high-security infectious disease unit for the care of such patients, with separate ventilation and waste-water treatment systems. The unit is also equipped to provide intensive care, and a laboratory can be rapidly set up and fully operative within 12-24 hours. Most important of all, personnel are available who are trained both for laboratory work and the care of such patients, and used to working as a team and familiar with the special protective equipment. lf a patient can not be transported to the special unit, a team is available to travel to the hospital where the patient has been admitted, to give instruction and help to set up infection control routines and even supply protective equipment.

Language: Swedish
Publication Type: Journal article, general information and opinions
Keywords: Infectious diseases / Haemorrhagic disease / Emergency services / Sweden

Fuller CC, Hartley B
Linear scleroderma: a Neuman nursing perspective
Journal of Pediatric Nursing 2000; 15(3): 168-174
NIVEL: C 8159

This article provides an overview of linear scleroderma (LS), a rare and chronic pediatric disease, which may leave scarring and may have devastating effects on a child and family. The authors present a case history of a boy with LS, and discuss how nurses and other health-team members can assist the child and family in managing the illness. The description of the nursing care is based on the Neuman systems model and on the nursing diagnoses of fear, disturbed self-concept, and impaired physical mobility.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Scleroderma / Children / Family / Nursing / Psycho social problems / Activities of daily living

Hennepe L te
Rare diseases need structured care: the diagnosis is just the start...
[Zeldzame aandoeningen vragen om gestructureerde zorg: de diagnose is nog maar een begin...]
Tijdschrift voor Huisartsgeneeskunde 1999; 16 (7/8): 374-376
NIVEL: C 8160

The author describes the problems and bottlenecks which are experienced by parents of children with rare diseases. Problems which occur are: lack of knowledge, a late diagnosis, which can have serious consequences, lack of continuity in care and psycho social problems, like stress within the family and lack of knowledge about where to get support. A multi disciplinary approach is necessary.

Language: Dutch
Publication Type: Journal article, general information and opinions
Keywords: Children / Parents / Continuity of care / Psycho social problems

Jansen PL
New criteria for liver transplantation in adults: the combined Groningen and Rotterdam protocol
The Netherlands Journal of Medicine 1998; 52(2): 53-57
NIVEL: C 8161

In this article a protocol for liver transplantation, written by the transplantation teams of Groningen and Rotterdam, is described. This is to ensure that the criteria for selection and timing of liver transplantation, and the procedures for patient evaluation, are identical or at least very similar. Also, the waiting list procedures in the two centres, including the criteria used to decide which patient will be transplanted first, are very similar. Currently, most patients with liver cirrhosis or filminant liver failure should be considered as liver transplant candidates unless a contraindication is found. Most contraindications are relative and are the subject of a careful balance of pros and cons. The detection and care of liver patients starts with the general practitioner. Referral to liver transplantation centres is the final step. In the Netherlands, liver disease is relatively rare and therefore patients are best served when the expertise is concentrated in a few liver centres.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Liver diseases / Liver transplantation / Practice guidelines / Coordination of care / Waiting lists / Patient selection

Kase BF, Nilsson B
A small-group centre: a model trial for activities of a centre for rare diseases and syndromes at the National Hospital
[Smågruppesenteret: en modellutprovning for Rikshospitalets utadrettede virksomhet for noen sjeldne sykdommer og syndromer]

Tidsskr Nor Laegeforen 1996; 116(11): 1340-1344
NIVEL: C 8162

In this article a description is given of the Centre for Rare Disorders at the National Hospital in Norway, a trial project in connection with the Government's Plan of Action for the disabled. The intention is to establish nationwide facilities which in cooperation with local resources can provide both medical, pedagogical and social services for persons with rare disorders and their families. The work at the centre is based on a life span perspective and the goal is improved coping, independence and better quality of life. The centre gathers, adapts and spreads information on 15 rare disorders. Representatives from the user organisations ensure that the users have a strong influence on the management of the centre. It is hoped that systematic evaluation of the centre's activities will give an answer to the Government and to the host hospital as to whether the intentions and goals will be fulfilled or not during the project period.

Language: Norwegian
Publication Type: Journal article, general information and opinions
Keywords: Community health services / Norway / Patient education / Social support

Keestra RW
Home visits to patients with an immunodeficiency
[Op huisbezoek bij patienten met een afweerstoornis]
Tijdschrift voor Kindergeneeskunde 1988; 56 (5): 218-225
NIVEL: C 8163

Objective: To gain insight into the problems that patients with a primary immunodeficiency (PID) experience at home. Primary immunodeficiencies are rare and therefore unknown not only to the broad public, but also to many doctors. Patients experience much uncertainty about the character of their ailment, the risk of infections, the treatment possibilities and the prognosis.
Methods: Patients with PID (N=87) and their general physicians were interviewed on their opinions and experiences with the illness and the treatment.
Results: The contact between PID patients and their general physician is, generally speaking, poor. Treatment is done mainly on a (poli)clinical basis, leaving the general physician out of the picture. Most patients say they would prefer home treatment; most general physicians say they would like to be more involved. The patient, however, is very often rushed to the hospital with the first symptoms of an infection.

Conclusions: It is argued to improve the contact between these patients and their general physicians, even when the disease requires treatment by (super)specialists. Considering the arguments it is a challenge for GP's and specialists to integrate both kinds of medical care, in order to prevent psycho social disorders as a sequel of a somatic disease. An organisation of PID patients is recommended.

Language: Dutch
Publication Type: Journal article, research
Keywords: Immunologic deficiency syndromes / Physician-patient relations / Family practice / Continuity of care / Patient organisations / Home care services / Patient satisfaction / Interview study

Kendrick D, Young A, Futers D.
The diagnosis and management of acute childhood illness: is there a role for health visitors?

Journal of Advanced Nursing 2000; 32(6): 1492-1498

Objective: To examine the current practice and attitudes of health visitors in identifying and managing acute childhood illness. Much of the published literature on nurse practitioners relates to nurses with backgrounds, training and nursing experience other than health visiting. Health visitors however may be ideally placed to develop their role in the identification and management of acute childhood illness, and such a development may benefit patients, the health service and health visitors themselves
Methods: A postal questionnaire survey was used in one community National Health Service (NHS) Trust.
Results: A total of 74 questionnaires were returned, a response rate of 55%. The majority of responding health visitors reported that they were already identifying and advising on a range of acute childhood illnesses. Their confidence in undertaking these activities varied by condition. They were less confident in identifying and advising on conditions requiring the use of diagnostic equipment, viral illnesses which are becoming increasingly rare, and those with potentially serious outcomes. More knowledge and more training were seen as methods for increasing confidence. Most health visitors could see advantages in taking on this role; almost two thirds felt it would become part of the role of the health visitor in the future and a high level of interest was expressed in developing such a role.
Conclusions: Many health visitors are already identifying and advising on a range of acute childhood illnesses and feel confident to do so. There may be advantages to the patient, the health service and the health visitor in

developing this role further, but such development would require specific training for health visitors.
Language: English
Document Type: Journal article, research
Keywords: Nurse practitioners / Primary health care / Children / Diagnosis / United Kingdom / Questionnaire study

Kumar H, Daykin J, Holder R, Watkinson JC, Sheppard MC, Franklyn JA
An audit of management of differentiated thyroid cancer in specialist and non-specialist clinic settings
Clinical Endocrinology (Oxf) 2001; 54(6): 719-723
NIVEL: C 8164

Objective: To audit the management of thyroid cancer, a rare endocrine malignancy, in a large university associated teaching hospital, to assess points of good practice identified from published guidelines and reviews, and to compare findings in groups of patients managed jointly by specialists with an interest in thyroid cancer (including surgeon, endocrinologist and oncologist) with a group managed by other clinicians outside that setting.
Methods: The case-notes of 205 patients with differentiated (papillary or follicular) cancer were reviewed retrospectively, including group A (N=134; managed in a specialist multi-disciplinary clinic setting) and group B (N=71; managed in other clinic settings). Points of good practice investigated were adequacy of surgery, surgical complications, prescription and adequacy of T4 treatment, adequacy of monitoring by measurement of serum thyroglobulin and action taken and appropriate administration of ablative radio iodine.
Results: Deficiencies in management of the cohort as a whole were identified, including inadequate surgery and inadequate TSH suppression in approximately one-fifth of the cases. Monitoring with thyroglobulin measurements and action when serum thyroglobulin was high were also inadequate in some cases and ablative radio iodine was not given, despite being indicated in 11.7% of the cohort. Inadequate surgery and failure to administer radio iodine were less common in those managed in a specialist clinic setting than in those managed in other clinic settings.
Conclusions: The findings highlight the need for locally agreed protocols in managing relatively rare endocrine disorders such as thyroid cancer and argue in favour of centralisation of expertise and patient management in multi-disciplinary specialist clinic settings.

Language: English

Publication Type: Journal article, research
Keywords: Thyroid cancer / Medical oncology / Quality of care / Multi disciplinary care / Medical audit / Treatment failure / Comparative study / Retrospective study

Lalatta F, Selicorni A, Briscioli V
Syndromes in outpatient experience
[Le síndromi nell'esperienza ambulatoriale]
La Pediatria Medica e Chirurgica 1993; 15 Suppl l: 60-63
NIVEL: C 8165

The authors report their experience as clinical geneticists and pediatricians in the management of patients affected by multiple congenital malformations and genetic syndromes. Their aim is to demonstrate that etiological diagnosis, crucial for successful management and counselling, has to be considered the first step in the care of these patients and their families. Coordination of medical interventions, clinical follow-up and psycho social support have to be included in every out-patient programme in this field. Diagnostic approach has to be carried out through a specific methodology which includes extensive clinical expertise in dealing with children affected by multiple congenital anomalies, protocols for major clinical problems, facilities for specific investigations (cytogenetic laboratory, molecular genetics, etc.), computerised programmes for recognising rare syndromes, network of motivated specialists (neurology, ophthalmology, orthopaedics, etc.) and possibility of collegial discussion of undiagnosed patients. Psycho social support and follow-up have to be achieved through trained professionals. Our 5 years of experience in this field (1420 patients) suggest the need for an organised network of services. This could provide the families of children affected by genetic syndromes with a comprehensive management and support programme which is lacking in the present health care system.

Language: Italian
Publication Type: Journal article, general information and opinions
Keywords: Genetic diseases / Children / Family / Coordination of care / Social support / Diagnosis

Mettlin C, Aungst CW, Michalek AM, Lane W, Gamble D
Comparison of patient profiles from a comprehensive cancer centre and the general population
Journal of Surgical Oncology 1984; 27(1): 8-11
NIVEL: C 8166

Objective: To get insight into the differences in cancer patient profiles between cases seen at comprehensive cancer centres and those seen in the general population. It was hypothesised that the patient population of a comprehensive cancer centre differs from the population seen in community hospitals, because of the near singular emphasis on cancer care, the patterns of referral and the nature of the research undertaken at such centres.
Methods: This report compares the characteristics of patients treated at Roswell Park Memorial lnstitute in Buffalo, a comprehensive cancer centre with all reported patients diagnosed in the Western New York population. Data for this study (resp. N=2005 and N=3066) were generated from the Roswell Park Tumour Registry and the Western New York Tumour Registry.
Results: The findings indicate that patients presenting for treatment at Roswell Park Memorial lnstitute, compared to the WNY cancer patient population, are more frequently diagnosed with rare tumours, are younger, and manifest later stage disease.
Conclusions: These data may have implications for the planning and evaluation of cancer patient services at the community level.

Language: English
Publication Type: Journal article, research
Keywords: Cancer / Cancer care facilities / Special hospitals / Registries / Comparative study

Rayhorn NJ, Ingebo KR
Aganglionosis of the small intestine: a rare form of Hirschsprung's disease
Gastroenterology Nursing 1999; 22(4): 164-166
NIVEL: C 8167

This article gives an overview of Hirschsprung's disease and presents three case studies of small bowel Hirschsprung's. Hirschsprung's disease is a condition found in newborns and associated with increased morbidity and mortality. Families are faced with numerous challenges, including maintenance of nutrition, the need for surgical interventions, and the potential for complications associated with the medical technologies necessary to maintain children with aganglionosis. Home care needs are not unlike those of other children with severe intestinal dysfunction such as short bowel syndrome. One centre’s experience with Hirschsprung's disease supports the need for a multi disciplinary approach to long-term management and home care.

Language: English

Publication Type: Journal article, general information and opinions
Keywords: Hirschsprung disease / Infants / Parents / Nursing / Home care services / Case report

Reinhold-Keller E, Herlyn K
Interdisciplinary management of vasculitis patients: internist / rheumatologist
[Interdisziplinäre Betreuung von Vaskulitis-Patienten: der Internist / Rheumatologe]
Zeitschrift fur Rheumatologie 2001; 60(4): 208-218
NIVEL: C 8168

This article gives an overview of the diagnosis and multi disciplinary management of systemic vasculitides (SV), a heterogeneous group of different entities with varying clinical and pathological-anatomical characteristics. At the onset of disease organ manifestations often present as a single symptom without appearance of indirect signs of vasculitides as musculoskeletal complaints and constitutional symptoms indicating inflammatory systemic disease. Therefore early interdisciplinary care is extremely important to avoid major organ involvement with the development of fatal disease. Besides the multi disciplinary physical examination serological and immunological parameters, particularly in small vessel vasculitides are relevant in establishing the diagnosis. Regarding the interdisciplinary care the authors differentiate between primary diagnostic procedures and continuous follow-up to observe therapeutic and side effects of medications. Instruments for the assessment of disease extent, activity and irreversible damage were developed in recent years to document prospectively the disease status and support activity-adjusted treatment. Because of the chronic relapsing character of systemic vasculitides, the measurement of health-related quality of life gained progressive interest in the longitudinal follow-up. In addition in these rare diseases early patient education with information on the disease, treatment, side effects and training in self management strategies will enable patients to actively participate in the management of their disease and bear responsibility.

Language: German
Publication Type: Journal article, review
Keywords: Systemic vasculitis / Diagnosis / Multi disciplinary care / Patient education / Quality of life

Storhaug K, Vandvik IH
Frambu Health Centre: promoting family focussed care for disabled children
International Journal of Rehabilitation Research 1983; 6(2): 175-182
NIVEL: C 8169

This article describes the activities of the Frambu Health Centre, which has evolved from a summer-camp site for children with poliomyelitis to a modern information and treatment Centre for families with disabled members. Since 1976, fortnightly courses have been held for an increasing number of patients with rare, often congenital and/or hereditary disorders (anorectal anomalies, bladder extrophy, congenital heart defects, cystic fibrosis, severe diabetes, haemophilia, hip joint defects, juvenile rheumatoid arthritis, minimal brain dysfunction, muscular dystrophy, phenylketonuria, psychosis/autism, spina bifida, Huntington's chorea, osteogenesis imperfecta, retinitis pigmentosa, a. o.). This article describes the facilities, operation, financing and staff at Frambu. An outline of the course programme is given. The contents of two research projects carried out at Frambu are described. When families with rare disorders meet for the first time, new perspectives open up. Exchange of experience and feelings, establishing lay organisations, collating and distributing information to professionals and families are some of the important results of the Frambu courses.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Children / Family / Community health centres / Social support / Patient education / Self-help groups

Tilli DJP, Terpstra SJ, Witte LP de
The role of a patient organisation in health care for the chronically ill and their families
First report: An evaluation of the activities of the Dutch Neuromuscular Diseases Association
[De rol van een patiëntenorganisatie in de zorg voor chronisch zieken en hun gezin
Eerste rapport: Een evaluatie van de activiteiten van de Vereniging Spierziekten Nederland]
Hoensbroek: Instituut voor Revalidatie-Vraagstukken, 1990
NIVEL: R 10226(1

Objective: To get insight into the participation and satisfaction of patients related to the activities of a Dutch patient organisation concerning muscular diseases.
Methods: A questionnaire was sent to 2031 members of the organisation, with a response of 59.1% (N=1169).
Results and conclusions: Major differences were found in participation in different activities. The magazine of the organisation is read by a great part of the members while discussion groups were attended by only a small part. Many patients reported not needing home visits or telephone contact. In general, the members are satisfied with the activities which are organised, especially concerning patient education and information supply.

Language: Dutch
Publication Type: Report
Keywords: Neuromuscular diseases / Patient organisations / Patient education / Social support / Patient satisfaction / Questionnaire study

Terpstra SJ, Witte LP de, Horst FGEM van der, Tilli DJP
The role of a patient organisation in health care for the chronically ill and their families
Second report: Living with a neuromuscular disease (I): an investigation under members of the Dutch Neuromuscular Diseases Association
[De rol van een patiëntenorganisatie in de zorg voor chronisch zieken en hun gezin
Tweede rapport: Leven met een neuromusculare aandoening (I): een onderzoek bij leden van de Vereniging Spierziekten Nederland]
Hoensbroek: Instituut voor Revalidatie-Vraagstukken, 1990
NIVEL: R 10226(2

Objective: To get insight into the impact of neuromuscular diseases on activities of daily living and quality of life. Besides, patient satisfaction, social support and participation in the activities of a Dutch patient organisation were investigated.
Methods: A questionnaire was sent to 2031 members of the organisation, with a response of 59.1% (N=1169).
Results and conclusions: The impact of neuromuscular disorders is especially high on physical functioning and less on psycho social functioning. A great part of the respondents reported being happy and satisfied. Problems are experienced in health care delivery, especially in a lack of coordination of care. Patient education activities by the patient organisation are experienced positively.

Language: Dutch
Publication Type: Report
Keywords: Neuromuscular diseases / Quality of life / Activities of daily living / Patient satisfaction / Patient organisations / Social support / Patient education / Questionnaire study

Terpstra SJ, Witte LP de, Horst FGEM van der, Tilli DJP
The role of a patient organisation in health care for the chronically ill and their families
Third report: Living with a neuromuscular disease (II): a comparison between members and non-members of the Dutch Neuromuscular Diseases Association
[De rol van een patiëntenorganisatie in de zorg voor chronisch zieken en hun gezin
Derde rapport:: Leven met een neuromusculare aandoening (II): een vergelijking tussen leden en niet-leden van de Vereniging Spierziekten Nederland]
Hoensbroek: Instituut voor Revalidatie-Vraagstukken, 1991
NIVEL: R 10226(3

Objective: To describe the impact of neuromuscular diseases on daily activities and the need for health care services, and to get insight into the participation in the activities of a Dutch patient organisation. The aim was to evaluate the activities of the organisation and their impact on quality of life.
Methods: A questionnaire was sent to 2031 members of the organisation and 81 patients who were not a member of the organisation. The response was 56% (N=1463).
Results and conclusions: Members reported more problems in physical functioning than non-members. No difference was found with respect to psycho social functioning and quality of life. More members had negative experiences with health care services than non-members. More members reported getting support than non-members. There are strong indications for a positive influence of membership of a patient organisation on quality of life.

Language: Dutch
Publication Type: Report
Keywords: Neuromuscular diseases / Patient organisations / Activities of daily living / Quality of life / Social support / Questionnaire study

Terpstra SJ, Witte LP de, Horst FGEM van der, Tilli DJP
The role of a patient organisation in health care for the chronically ill and their families
Fourth report: Health care for patients suffering a neuromuscular disease: the development of a health care model
[De rol van een patiëntenorganisatie in de zorg voor chronisch zieken en hun gezin
Vierde rapport:: De zorg voor patiënten met een neuromusculare aandoening: de ontwikkeling van een zorgmodel]
Hoensbroek: Instituut voor Revalidatie-Vraagstukken, 1992
NIVEL: R 10226(4

Objective: To investigate the influence of the activities of a Dutch patient organisation concerning neuromuscular diseases on health care services and health care utilisation.
Methods: Hundred respondents of a former investigation were selected (50 members of the patient organisation and 50 non-members), who were interviewed.
Results and conclusions: Members make less frequently use of professional care than non-members. No differences were found between members and non-members with respect to coordination of care. Both groups report that an important role for the patient organisation lies in patient education and, for a smaller group, offering social support. A proposal is given for a health care model in which coordination of care plays an important part.

Language: Dutch
Publication Type: Report
Keywords: Neuromuscular diseases / Patient organisations / Health care utilisation / Coordination of care / Patient education / Social support / Interview study

Truckenbrodt H
The treatment organisation of rheumatic children and adolescents in West Germany
[Zur Versorgungsstruktur rheumakranker Kinder and Jugendlicher in Westdeutschland]
Zeitschrift für Rheumatologie 1990; 49(5) : 260-267
NIVEL: C 8170

This article addresses the issue of pediatric rheumatology, a specialised subject which overlaps with important fields of pediatrics, as well as with adult rheumatology. As a speciality it includes age-specific rheumatologic diseases, an age-dependent differential diagnosis, special growth

disturbances and joint deformities that are also influenced by age, and a subgroup of juvenile chronic arthritis. Socio-medical problems arise from a child's impaired physical and psycho social development and their impact on education, the individual's choice of career, and effects on the patient's family. For pediatricians who are trained in this field and who treat only or predominantly children with rheumatic diseases the subspeciality of pediatric rheumatology should be introduced. In the future, precise figures on incidence and prevalence of juvenile rheumatic diseases in Germany must be obtained. Therapy of children with chronic arthritis affords improvement and further development by comprehensive training and further education of physicians and physiotherapists. Thus, it is important to take advantage of possibilities of inpatient and outpatient care, as well as the coordination of existing institutions. Arthralgias and acute rheumatic disorders in children present in a relatively high frequency. Chronic arthritis, however, is rare and treatment requires a supra-regional centre for pediatric rheumatology. Quality of therapy for children with chronic rheumatic diseases must be considered prior to a locally oriented care. More intensive research is needed, with more clinical studies and basic research. More effort must be made to integrate analyses of musculoskeletal function and the importance of joint-deformity development into the research programme. Further cooperation between pediatric rheumatology centres and universities would offer significant advantages in the future.

Language: German
Publication Type: Journal article, general information and opinions
Keywords: Rheumatoid arthritis / Children / Coordination of care / Epidemiology / Organisational structure / Research / Germany

Vandvik IH, Storhaug K
Family-focussed services for children with rare disorders, exemplified by bladder exstrophy: a Norwegian national health programme
Clinical Pediatrics (Phila) 1985; 24(2): 97-100
NIVEL: C 8171

This paper describes the Frambu Health Centre, which offers family-focussed services to patients with bladder exstrophy during the first information and treatment course arranged for this group. Main areas of concern as reported by the families are presented, like feelings of shame, lack of social support and the need for more information. The services of the Frambu Health Centre include lectures, private consultations and group discussions, with the aim to make families better equipped and informed to look after their disabled members.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Bladder extrophy / Children / Family / Community health centres / Social support / Patient education / Psycho social problems / Self-help groups

2.2.2 Relationship between patient and health care professional

Atherton A
Primary care for patients with care chronic illnesses
European Journal of General Practice 1997; 3: 58-61

Objective: To investigate the experiences, needs and expectations of primary care reported by patients with rare chronic illness.
Methods: Randomly selected parents of children with one rare chronic condition were interviewed (N=18).
Results: Information was the most frequently expressed need, but patients were realistic about what the general physician could provide, and paradoxically, honesty about a lack of knowledge was admired. Willingness to help with seeking information was positively perceived. Patient reporting of the morbidity due to rare illnesses was often discounted. Parents expressed satisfaction with general physicians who supported them in using alternative treatments. The parents correctly understood that the common illnesses were a priority issue, but felt themselves to be marginalised.
Conclusions: The needs of patients with rare chronic illnesses in general practice should be further explored.

Language: English
Publication Type: Journal article, research
Keywords: Chronic diseases / Children / Primary care / Information need / Physician-patient relations / Interview study

Boyd JR
A process for delivering bad news: supporting families when a child is diagnosed
Journal of Neuroscience Nursing 2001; 33(1): 14-20
NIVEL: C 8172

This article describes a process for delivering bad news to families of children with a neurodegenerative disorder to enhance the families' coping abilities and to lessen the impact of the devastating news. The description of this process is guided by a review of the literature on parental satisfaction with and preferences for diagnosis delivery, published guidelines for

breaking bad news, and the concepts of coping, grief, support, hope, and family-centred care. Nurses play a collaborative role in the implementation of this process that remains sensitive to the needs of each family. Their influence on how the interaction takes place and their support of the family both during and after they receive bad news are pivotal. Research to formally evaluate the outcomes of the interventions and strategies within the process is necessary.

Language: English.
Publication Type: Journal article, review
Keywords: Neurodegenerative diseases / Truth disclosure / Professional-family relations / Children / Coping / Social support / Grief

Fang-Kircher S, Kraft M
Mucopolysaccharidoses from the view point of mucopolysaccharidoses families: 10 years experiences of the "Austrian Society for Mucopolysaccharidoses"
[Die Mukopolysaccharidosen aus der Sicht der MPS-Familien: 10 Jahre Erfahrungen der "Österreichischen Gesellschaft für Mukopoly-saccharidosen"]
Wiener Klinische Wochenschrift 1996; 108(2): 29-32
NIVEL: C 8173

In this article a rare inborn disorder of metabolism, called mucopolysaccharidoses (MPS) is described. The disorder is caused by defects in enzymes which are necessary for the degradation of mucopolysaccharides. An effective causal treatment is not available as yet. Next the authors give their opinion about the role of the doctor, who, both from the medical and human aspect, has to assist MPS patients and their families physically and psychologically over many years. This task needs much empathy, working morale and knowledge of these diseases on the part of the medical adviser involved, but also demonstrates the limitations of active medical treatment. Since 10 years the "Austrian Society for Mucopolysaccharidoses" has tried to fill the gap between the MPS families' need for help and the still discouraging current medical treatment prospectives.

Language: German
Publication Type: Journal article, general information and opinions
Keywords: Mucopolysaccharidoses / Children / Family / Psycho social problems / Social support / Patient organisations

Hofmans-Okkes IM
Patients with rare and serious chronic diseases in primary care: a blind spot in the literature
[Patiënten met een zeldzame, ernstige chronische ziekte in de huisartspraktijk: een witte vlek in de literatuur] Huisarts en Wetenschap 1996; 39 (7): 316-323

Objective: To get insight into the (problems in the) relationship between general physicians and patients with rare, serious chronic diseases.
Methods: A literature search was performed to several rare diseases in combination with terms expressing the concepts of primary care and patient perspective. The literature was ordered according to a model in which the essential aspects of the contact between patient and physician are outlined.
Results: As an example, the specific results are given for cystic fibrosis. Study books, journal articles and information materials from the patient organisation were found.
Conclusions: There is a large literature on rare chronic diseases: however, through this study's search, relatively little factual information was found on (problems in) the contact between general physician and patients with these diseases. The literature mostly originates from the orbit of medical specialists and is highly disease oriented. A study is recommended into what actually occurs in this field, aiming at collecting and describing the experience and know-how of both general physicians and patients in order to develop a strategy for the contact between general physician and these patients (and their families).

Language: Dutch
Publication Type: Journal article, literature review
Keywords: Chronic diseases / Cystic fibrosis / Primary care / Physician-patient relations / Literature study

Hofmans-Okkes IM, Meyboom-de Jong B, Lamberts H, Weel C van
Chronically ill patients with rare diseases in family practice
[Chronisch zieken met zeldzame ziekten in de huisartspraktijk]
Zoetermeer: Nationale Commissie Chronisch Zieken, 1994
NIVEL: B 3571

Objective: To get insight into what general physicians contribute to the care of patients with rare chronic diseases, and into the problems which are experienced both by patients and general physicians.
Methods: A matrix model was developed to describe the different aspects of the relationship between general physician and patient. A literature study

was performed in order to fill in the matrix for 10 rare diseases. To compare the amount of available information with the information about more frequent disorders a study is also made of the literature concerning cerebrovascular accident. This study was performed under the auspices of a Dutch national committee on the care for chronically ill patients.
Results: The results indicate that many problems are experienced in the relationship between general physician and patient. There is not much literature about the prevalence of rare diseases in family practice, nor about the actions undertaken by general physicians.
Conclusions: There seems to be a shortage of expertise on rare diseases in family practice. Specific post-graduate courses about rare diseases do not seem an appropriate solution, because there exist more than 5000 rare diseases. But there is a need for further research and different methods are suggested.

Language: Dutch
Publication Type: Research report
Keywords: Multiple sclerosis / Psoriasis / Phenylketonuria / Chronic fatigue syndrome / Schizophrenia / Cystic fibrosis / Spondylitis / Posttraumatic dystrophy / Macular degeneration / Liver cirrhosis / Liver transplantation / Cerebrovascular accident / Family practice / Physician-patient relations / Literature study

Nishio KT
A family affected by mitochondrial encephalomyopathy: a nursing protocol
Clinical Nurse Specialist 1997; 11(5): 195-201
NIVEL: C 8174

The purpose of this article is twofold: (1) to provide an overview of mitochondrial encephalomyopathies, rare genetic diseases that affect organs and tissues with high oxidative activity, such as the brain, striated muscles, and heart, and (2) to present a protocol to guide nursing practice. Protocol development is based on a case history and guided by Dungan's model of dynamic integration. The protocol addresses perceived family needs, nursing interventions, and outcomes of care.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Mitochondrial encephalomyopathies / Genetic diseases / Children / Nursing / Guidelines / Case report

Starke M, Wikland KA, Moller A
Parents' experiences of receiving the diagnosis of Turner syndrome: an explorative and retrospective study
Patient Education and Counseling 2002; 47(4): 347-354

Objective: To explore the experiences of parents whose children are diagnosed of having Turner syndrome (TS), a sex-chromosome disorder occurring in approximately 1:2500 female births.
Methods: In this explorative and retrospective study, parents (33 mothers and 11 fathers) from different families, each containing a girl with TS, were interviewed to explore their perceptions and experiences of the diagnostic process, including the period of investigation and the time when the diagnosis was given. There was wide variation in degree of retrospection in this study; one parent who was interviewed had a daughter of 17 years of age who was diagnosed before 1 month of age. On the other hand, two girls had been diagnosed only 6 months before their parents were interviewed.
Results: Most parents (N=27) reported that it was professionals who observed that their daughter should be referred for investigation, and the remainder (N=17) reported that they themselves perceived the need for investigation. Their experiences were found to affect the parents' descriptions of the diagnostic process. The parents who initiated the investigation themselves described the diagnosis in terms of confirmation of their own suspicions. When professionals initiated the investigation the parents reported that they felt unprepared when they received the diagnosis.
Conclusions: The results of this study indicate that the parents' experiences of receiving the diagnosis of TS appear to depend on the ability of the doctor to provide relevant information and to manage the psycho social aspects correctly. A better understanding of parents' experiences should enable health professionals to counsel parents more effectively concerning the diagnosis of TS.

Language: English
Publication Type: Journal article, research
Keywords: Turner syndrome / Children / Parents / Diagnosis / Professional-patient relations / Interview study

2.2.3 Information and patient education

Eboh WO, Akker O van den
Service provision for sickle-cell disease: school-based education
British Journal of Nursing 1996; 5(1): 30-33
NIVEL: C 8176

In this article it is argued that school children should receive instruction about the possible risks and effects of sickle-cell disorders as part of a health education programme, to enable them to make informed decisions about their future. With the establishment of multicultural societies it is increasingly likely that many people will be in contact with people affected by the disease. The general lack of understanding has led to many misjudgements being made about people's health status. In this article the objectives and issues of school health education programmes are described.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Sickle cell disease / Children / Patient education / School health services

Ham M van der, Hennepe L te
Health education about rare hereditary diseases: a study of health care needs in patient organisations
[Voorlichting over zeldzaam voorkomende aangeboren en/of erfelijke aandoeningen: een behoeftenpeiling bij de lidorganisaties van de VSOP]
Soestdijk: VSOP, 1998
NIVEL: R 10291

Objective: To get a clear picture of which kinds of patient education materials on rare hereditary disorders are needed by Dutch patient organisations.
Methods: Key figures of 11 patient organisations filled in a questionnaire about information needs. The same questionnaire was filled out by some members of the information service of the Dutch coordinating organisation of patient organisations (VSOP).
Results: Only 5 organisations expressed their need for patient education materials. Descriptions of several rare diseases themselves were needed. Criteria were determined for which diseases a description could be made in order to make an information leaflet.
Conclusions: A possible explanation for the low number of organisations expressing their need for patient education materials could lie in the fact that there is not a solid relationship between patient organisations and the coordinating organisation. A more active role of the coordinating organisation is recommended.

Language: Dutch
Publication Type: Report
Keywords: Genetic diseases / Patient organisation / Information need / Information supply / Questionnaire study

Soumerai SB, Ross-Degnan D, Kahn JS
Effects of professional and media warnings about the association between aspirin use in children and Reye's syndrome
The Milbank Quarterly 1992; 70(1):155-182

This article addresses the question of how the media can change consumer behaviour regarding popular health-related products by focussing on the diffusion of risk information about the relationship between the use of aspirin and Reye's syndrome, a rare but potentially fatal or severely debilitating illness, which usually occurs in children with flu or other viruses. First, the epidemiological evidence is reviewed. Second, the actions of several public and private organisations and the key events that promote or impede public awareness are described. Third, the authors report on the relationship between, on the one hand, the quantity and timing of medical and lay media attention, combined with other public education activities, and, on the other hand, decreases in the use of aspirin for children and reductions in disease incidence. Finally, some characteristics of risk communication are proposed that may facilitate changes in health behaviours through the use of mass communications.

Language: English
Publication Type: Journal article, review
Keywords: Reye's syndrome / Children / Aspirin / Patient education / Mass media / Risk communication

VSOP
Information services for patients suffering rare diseases: a study of available information, problems and solutions
[Informatie voor mensen met een zeldzame aandoening: een onderzoek naar de beschikbare informatie, knelpunten en oplossingen]
Soestdijk: VSOP, 2002
NIVEL: R 10292

Objective: To improve the information supply on rare diseases for patients and their relatives.
Methods: Questionnaires were sent to patient organisations (N=70), and to patients or parents of children suffering a rare disease (N=400). The questionnaire consisted of items concerning the supply of information by health care professionals and the availability of information. Six policymakers and board members were interviewed to get an impression of their policy plans.

Results: Of the 70 questionnaires to the patient organisations, 44 were received (63%). 35,5% (N=167) of the patients and parents filled out the questionnaire. The disease was diagnosed by a specialist and frequently this took more than 2 years. Information about diseases was mostly given by patient organisations. The disease had a high impact on daily life. Psycho social support was not available sufficiently. Patients had a lack of knowledge about treatment possibilities. The interviews with policymakers resulted in a clear impression of the bottlenecks in the care and treatment of rare diseases.
Conclusions: Recommendations for improvement in the diagnostic process, care, treatment, information supply and policy are given. Early diagnosis, research and information supply should be stimulated.

Language: Dutch
Publication Type: Report
Keywords: Information supply / Quality improvement / Diagnosis / Orphan drugs / Health care services / Public policy / Questionnaire study

2.3 Outcome

2.3.1. Quality of care

Bernstein RM, Hollingworth GR, Wood WE
Prompting physicians for cost-effective test ordering in the low prevalence conditions of family medicine
Proceedings of the Annual Symposium on Computer Application in Medical Care 1994: 824-828
NIVEL: C 8177

This article offers a description of a computerised prompting system for test ordering which is developed both in order to decrease the cost of investigations and to promote an evidence based learning approach to test ordering. Prompting systems have been shown to be cost-effective but suffer from many disadvantages in the family practice setting. They tend to be difficult to modify by the user and contingent on an inflexible rule based structure. Many suggestions are ignored implying that they are not relevant. In family practice most conditions are of low prevalence. Prompting for test ordering where the pre-test likelihood of disease is small will result in a large number of false positives and many unnecessary repeat or confirmatory investigations and attendant anxiety unless the prompting system is specifically designed to be used in a low prevalence environment. PROMPTOR-FM (PRObabilistic Method of Prompting for Test ORdering in

Family Medicine) was developed to overcome these perceived difficulties. It allows the physician to rapidly calculate the positive and negative predictive values of a test being considered based on the clinical index of suspicion. The physician is able to repeat the calculations and compare the results with previous calculations. By using PROMPTOR-FM repetitively, the clinician can learn to balance the risk of "missing" a rare but serious condition against the risk of falsely identifying disease with its downstream hazards and costs of further investigation. Prompting for test ordering is therefore uniquely tailored to each patient's situation.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Diagnosis / Medical informatics / Evidence based medicine / Primary care

Blakely ML, Lally KP
Using evidence-based medicine principles to improve quality of patient care in pediatric surgery
Seminars in Pediatric Surgery 2002; 11(1): 42-45
NIVEL: C 8178

Principles of Evidence Based Medicine (EBM), with the goal of improving patient care, are outlined, including a hierarchy of levels of evidence. The obstacles of these principles in pediatric surgery are discussed, especially the problem in conducting EBM studies because of the rarity of many disease conditions. However, with careful planning and funding, such studies can and should be performed, and the ways how quality of care can be improved are discussed.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Evidence based medicine / Surgery / Pediatrics / Quality of care

Co JP, Johnson KB, Duggan AK, Casella JF, Wilson M
Does a clinical pathway improve the quality of care for sickle cell anaemia?
Joint Commission Journal on Quality and Safety 2003; 29(4): 181-190

Objective: To describe how quality of care and outcomes change after introduction of a clinical pathway to improve the care for pediatric sickle cell vaso-occlusive crisis (VOC) .
Methods: A restrospective study, before and after introduction of the clinical pathway.

Results: Physicians used the pathway in 43% of eligible admissions, with use decreasing over time. Patients on the pathway were more likely to receive each of its required elements than those not on the pathway (odds ratios [OR] 1.15-2.49). After pathway implementation, even patients not on the pathway were more likely to receive incentive spirometry than those admitted before pathway availability (OR 1.40). Pathway use was associated with longer length of stay (LOS) and time to oral pain medication, while readmission rates did not change.
Conclusions: Use of a clinical pathway improved quality of care by increasing compliance with specific care elements, with mixed results on outcomes. Pathways may improve care for all patients, including nonpathway-treated patients, by influencing underlying practice patterns. Quality improvement committees must regularly monitor outcomes after pathway implementation to evaluate the need for pathway reinforcement and refinement.

Language: English
Publication Type: Journal article, research
Keywords: Sickle cell anaemia / Quality of care / Guidelines / Evidence based medicine

Collette L, Sylvester RJ, Stenning SP, Fossa SD, Mead GM, de Wit R, de Mulder PH, Neymark N, Lallemand E, Kaye SB
Impact of the treating institution on survival of patients with "poor-prognosis" metastatic nonseminoma European Organisation for Research and Treatment of Cancer Genito-Urinary Tract Cancer Collaborative Group and the Medical Research Council Testicular Cancer Working Party
Journal of the National Cancer Institute 1999; 91(10): 839-946
NIVEL: C 8146

Objective: To explore whether there is an association between the experience of the treating institution with respect to rnetastatic nonseminomatous germ cell cancer, a rare but treatable cancer, and the long-term clinical outcome of the patients, particularly patients with a poor prognosis.
Methods: Data were analysed on 380 patients treated in one of 49 institutions participating in the European Organisation for Research and Treatment of Cancer / Medical Research Council randomised trial of four cycles of bleomycin-etoposide-cisplatin followed by two cycles of etoposide-cisplatin versus three cycles of bleomycin-vincristine-cisplatin followed by three cycles of etoposide-ifosfamide-cisplatin-bleomycin, both treatment regimens given with or without filgrastim (granulocyte colony-

stimulating factor). Institutions were divided into four groups based on the total number of patients entered in the trial. The groups were compared by use of the Cox proportional hazards model stratified for treatment with filgrastim and for patient prognosis as defined by the International Germ Cell Consensus Classification Group. With the use of this classification, only 65% of the patients had a poor prognosis.
Results: Patients treated in the 26 institutions that entered fewer than five patients into the trial had an overall survival that was statistically significantly worse (two-sided P = .010; hazard ratio = 1.85; 95% confidence interval = 1.16-3.03) than that of patients treated in the 23 institutions that entered five patients or more. Overall survival and failure free survival were similar among institutions that entered at least five patients. The observed effect may be related to differences in adherence to the chemotherapy protocol and in the frequency and extent of surgery for residual masses, although only the differences in dose intensity achieved statistical significance.
Conclusions: Patients treated in institutions that entered fewer than five patients into the trial appeared to have poorer survival than those treated in institutions that entered a larger number of patients with "poor prognosis" nonseminoma.

Language: English
Publication Type: Journal article, research
Keywords: Germinoma / Chemotherapy / Outcome assessment / Disease-free survival / Professional competence

Eisen A
How to improve the diagnostic process
Journal of Neurology 1999; 246 Suppl 3: III6-9
NIVEL: C 8179

This article deals with the difficulties in the diagnostic process of amyotrophic lateral sclerosis (ALS). Although the existing therapeutic options for treatment are relatively limited, there will be a need for early diagnosis as treatment possibilities of this rare but fatal neurodegenerative disorder are improving. Early diagnosis not only requires the utility of a good biological marker, it also requires medical personnel who are experienced to consider/establish a diagnosis of ALS. In many countries, diagnosis of ALS follows a three-step process involving the family physician/practitioners, a community neurologist and an ALS specialist. This process is cumbersome and as the number of cases seen by the first two doctors is extremely small, their awareness is consequently low. There is an

urgent need to improve the awareness of ALS in the general physicians but also within the general population. A number of elements which probably contribute to delays in achieving an early diagnosis of ALS are discussed with the view of identifying how the diagnostic process can be improved.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Amyotrophic lateral sclerosis / Diagnosis / Quality of care

Jason LA, Richman JA, Friedberg F, Wagner L, Taylor R, Jordan KM
Politics, science, and the emergence of a new disease: the case of chronic fatigue syndrome
American Psychologist 1997; 52(9): 973-983
NIVEL: C 8150

This article addresses the controversy about chronic fatigue syndrome (CFS), which emerged as a diagnostic category during the last decade. Initial research suggested that CFS was a relatively rare disorder with a high level of psychiatric comorbidity. Many physicians minimised the seriousness of this disorder and also interpreted the syndrome as being equivalent to a psychiatric disorder. These attitudes had negative consequences for the treatment of CFS. By the mid-1990s, findings from more representative epidemiological studies indicated considerably higher CFS prevalence rates. However, the use of the revised CFS case definition might have produced heterogeneous patient groups, possibly including some patients with pure psychiatric disorders. Social scientists have the expertise to more precisely define this syndrome and to develop appropriate and sensitive research strategies for understanding this disease.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Chronic fatigue syndrome / Epidemiology / Mental disorders / Diagnosis / Diagnostic errors / Therapeutics / Research

Overgaag A, Oudenampsen D
Problems in health care services for people suffering postpoliomyelitis syndrome
[Knelpunten in de zorg voor mensen met het postpoliosyndroom]
Utrecht: Verwey-Jonker Instituut, 2001
NIVEL: R 10294

Objective: To get insight into the problems which are experienced by patients suffering postpoliomyelitis syndrome and to give advise about improving quality of care.
Methods: A questionnaire was constructed, based on a list of quality criteria. This questionnaire was filled in by 280 patients who were members of a Dutch patient organisation concerning neuromuscular diseases.
Results and conclusions: Problems were reported with respect to referral by general physicians to specialists, recognition and diagnosis of the disease, and continuity and coordination of care. Many patients were not satisfied with the way they were treated by general physicians, neurologists or company doctors. Quality of care has to be improved with respect to these problems, for which advice is given by expert groups.

Language: Dutch
Publication Type: Report
Keywords: Postpoliomyelitis syndrome / Quality of care / Patient satisfaction / Continuity of care / Patient education

Pollemans MC, Leeuwen YD van, Mol SSL, Grol R, Schadé E, Tan LHC
General practitioners' knowledge about chronic diseases
[Kennis van huisartsen over chronische aandoeningen]
Huisarts en Wetenschap 1996; 39 (7): 328-331

Objective: To get insight into general practitioners' (GPs) knowledge on chronic diseases.
Methods: A test on general practice was filled in by 906 GPs or GPs in training (N=906). This test included 160 case oriented items, of which 30 concerned chronic diseases.
Results: The highest scores were obtained by recently qualified GPs. The more experienced GPs were, the lower their scores. As far as knowledge on chronic diseases is concerned, this is obtained during the later stages of education and remains stable during the career. A relationship was found between practical experience and knowledge.
Conclusions: Because knowledge on certain chronic diseases depends on practical experience, i.e. how often a GP gets confronted with them, there is a need of greater patient variation in the practical stages of education, training and postgraduate courses. This is especially important for those diseases and problems which are not often seen in practice, like rare diseases.

Language: Dutch

Publication Type: Journal article, research
Keywords: Chronic diseases / Family practice / General practitioners / Professional competence / Medical education / Continuing education / Questionnaire study

Ratner V
Current controversies that adversely affect interstitial cystitis patients
Urology 2001; 57(6 Suppl 1): 89-94
NIVEL: C 8180

This article offers a description of certain areas of controversy in the field of lnterstitial Cystitis research. Interstitial Cystitis (IC) is a rare disease, characterised by pelvic pain, urinary urgency and frequency, which can dramatically affect quality of life. Many physicians still do not believe that IC exists, or else believe that it is a rare postmenopausal condition. This can cause significant delays in diagnosis and treatment. It is particularly problematic in children, whose symptoms are often diagnosed as "voiding dysfunction" and are thought to be self-limiting. It can also be problematic for men, who are often unsuccessfully treated for prostatitis over the course of many years, and for whom the diagnosis of IC is never entertained. In some cases, when no diagnosis is made, patients are left to live with severe, debilitating symptoms and have nowhere to turn for help. Resistance to treating severe nonmalignant pain with opioid medication further compounds this problem and has led to suicide in this patient population. The "gold standard" of cystoscopy with hydrodistention is now being questioned, and an over reliance on the potassium test, which has a high false-negative rate, may lead to significant underdiagnosis of IC. New urinary markers hold promise for both diagnostic as well as therapeutic potential, but are not yet commercially available. IC may be an organ-specific disease in some patients and a systemic condition in others. Many patients have multiple disorders and have no physician to manage their overall health. The lnterstitial Cystitis Association believes that the best way to address these unresolved areas of knowledge is to: (1) educate patients on all available diagnostic and therapeutic options so that patients, with the help of their physicians, can make the best informed decisions possible, and (2) aggressively pursue all avenues of research, particularly epidemiology.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Interstitial Cystitis / Diagnosis / Diagnostic errors / Quality of life / Quality of care / Patient organisations / Patient education

Sacchetti A, Sacchetti C, Carraccio C, Gerardi M
The potential for errors in children with special health care needs
Academic Emergency Medicine 2000; 7(11): 1330-1333
NIVEL: C 8154

This article addresses potential sources of errors in the management of children with special health care needs. These children are at risk for suboptimal treatment when presenting for emergent care to unfamiliar health care providers. Errors in their management may stem from failure to recognise occult conditions, lack of familiarity with rare or complex medical problems, or lack of prior knowledge of baseline physical findings. An emergency information form (EIF) that contains patient-specific information on essential diagnostic and therapeutic interventions may provide a ready personal reference for the emergent care of these children. Coupled with the use of medical identification jewelry and an electronic transmission system, an EIF has the potential to eliminate management errors in the care of these patients.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Children / Diagnosis / Medical errors / Emergency services / Medical records

Simpson DE, Rich EC, Dalgaard KA, Gjerdingen D, Crowson TW, O'Brien DK, Johnson PE
The diagnostic process in primary care: a comparison of general internists and family physicians
Social Science & Medicine 1987; 25(7): 861-866
NIVEL: C 8155

Objective: To get insight into the formulation of diagnostic hypotheses by general internists and family physicians in response to three patient cases (dyspnea, abdominal pain and syncope).
Methods: The investigation was conducted in the United States. The responses of 24 primary care physicians (N=24), 12 internists (N=12) and 12 general physicians (N=12) to sequentially presented written clinical information were audiotaped. Each transcribed protocol was scored to enumerate and characterise the hypotheses considered by physicians in each specialty.
Results: Results of the analyses of variance of hypothesis measures revealed that internists generated more hypotheses than general physicians and that the internists' hypotheses were more specific and were less likely to be generated by other physicians. In addition, internists tended to consider

hypotheses more closely related to the final diagnosis sooner in the case presentation than did general physicians.
Conclusions: The findings of increased number, specificity, and uniqueness of hypothesis considered by internists are consistent with previously demonstrated differences in the amount and nature of diagnostic information collected by general physicians and internists. The results suggest that internists have additional experience-based expertise in the diagnosis of less common problems.

Language: English
Publication Type: Journal article, research
Keywords: Diagnostic techniques and procedures / General physicians / Internists / Professional competence / Comparative study

Spickett GP, Askew T, Chapel HM
Management of primary antibody deficiency by consultant immunologists in the United Kingdom: a paradigm for other rare diseases
Quality in Health Care 1995; 4(4): 263-268
NIVEL: C 8181

Objective: To get insight into the variation in clinical practice and its effect on outcome for rare diseases such as primary antibody deficiency.
Methods: As part of a national audit a survey of all consultant immunologists (N=30) in the United Kingdom dealing with primary antibody deficiency syndromes in adults and children was carried out in 1993 to ascertain their practices in diagnosis and management. Consensus guidelines were published after the survey was completed.
Results: Comparison of the survey results of clinical practice at the time the guidelines were published with the standards identified highlighted that the practice of a minority of specialists was at variance with their peers and with the consensus document, particularly in the use of intramuscular immunoglobulin, the dose and frequency of intravenous immunoglobulin, and target through immunoglobulin G concentration, which has implications for the quality of patient care. However, much closer agreement existed in the key areas of management, such as diagnosis and selection of intravenous immunoglobulin.
Conclusions: The approach and the problems identified are relevant to the management of other rare diseases, in which diagnosis and management is complex and there are few specialists with the necessary knowledge to undertake such care. This survey, the first attempted audit of practice, shows that within a motivated group of specialists highly significant differences in

practice may exist and the authors emphasise the importance of setting clear guidelines against which care can be assessed.

Language: English
Publication Type: journal article, research
Keywords: Primary antibody deficiency syndromes / Guidelines / Survey

2.3.2 Quality of life

Aronson KJ
Quality of life among persons with multiple sclerosis and their caregivers
Neurology 1997; 48(1): 74-80
NIVEL: C 8182
Objective: To get insight into the quality of life (QOL) among persons with multiple sclerosis (MS) and their caregivers and into the psycho social consequences of this illness.
Methods: A large population-based survey of persons with MS and their caregivers (N=11752) in Ontario was conducted using self-completed mailed questionnaires. The objectives included describing satisfaction with QOL and determining relationships between QOL as a whole and several other factors, such as demographic characteristics and measures of physical disability.
Results: Response rates were 83% for those with MS and 72% for their caregivers. Based on 697 respondents with MS, mean age was 48 years, 70% were women, and 75% were married. While 24% experienced no mobility restrictions, the majority required some type of aid or a wheelchair for getting around. Health received the lowest satisfaction rating among the six components of QOL, while finances received a relatively low satisfaction rating from the 345 caregivers.
Conclusions: Less satisfaction with several QOL components was evident for those with MS compared with the disabled in the Canadian general population, and for caregivers compared with the able-bodied general population. Poorer QOL as a whole among those with MS was associated with unemployment, MS symptoms of moderate or worse, fatigue, mobility limitations on stairs, a disease course other than stable, and was most strongly related to interference by MS in social activities. Among caregivers, poorer QOL as a whole was associated with being a spouse, longer duration of care giving, moderate or worse MS symptoms in the care recipient, and most strongly related to a care recipient's current MS disease course of other than stable. Through an understanding of the satisfaction with QOL of persons with MS and caregivers, and the relationships with other important

factors, autonomy and home care may be supported and prolonged, while preventing unnecessary institutionalisation.

Language: English
Publication Type: Journal article, research
Keywords: Multiple sclerosis / Quality of life / Physical disability / Patients / Caregivers / Psycho social problems / Survey

Benrud-Larson LM, Haythornthwaite JA, Heinberg LJ, Boling C, Reed J, White B, Wigley FM
The impact of pain and symptoms of depression in scleroderma
Pain 2002; 95(3): 267-275
NIVEL: C 8185

Objective: To determine the frequency and impact of pain, symptoms of depression, and social network characteristics on physical functioning and social adjustment in patients with scleroderma, a rare connective tissue disease that can affect multiple organ systems.
Methods: Scleroderma patients (N=142) completed measures of pain, depressive symptoms, social network characteristics, physical functioning, and social adjustment.
Results: Sixty-three percent reported at least mild pain and 50% reported at least mild levels of depressive symptomatology. Hierarchical regression analyses revealed that pain, depressive symptoms, and employment status (disabled/unemployed vs. not) were significant, independent predictors of physical functioning, together accounting for 37% of the total variance. Pain was the single strongest predictor of physical function, accounting for 20% of the variance. Depressive symptoms, physical functioning, diversity of social network, and employment status were significant independent predictors of social adjustment, together accounting for 63% of the variance. Depressive symptoms were the single strongest predictor of social adjustment, accounting for 26% of the variance. The effects of pain and physical function on social adjustment became non-significant when depressive symptoms were entered into the model, suggesting that symptoms of depression mediate the effect of pain and physical functioning on social adjustment.
Conclusions: These findings indicate that pain is common in scleroderma and that pain and depressive symptoms are significant determinants of physical functioning and social adjustment, two important components of health-related quality of life. Increased attention to effective management of pain and symptoms of depression in scleroderma will likely lead to improved functioning and quality of life.

Language: English
Publication Type: Journal article, research
Keywords: Scleroderma / Pain / Depression / Social support / Social adjustment / Employment / Disability / Questionnaire study

Green PHR, Stavropoulos SN, Panagi SG, Goldstein SL, Mcmahon DJ, Absan H, Neugut AI
Characteristics of adult celiac disease in the USA: results of a national survey
American Journal of Gastroenterology 2001; 96(1): 126-131
NIVEL: C 8183

Objective: To get information about the clinical spectrum of adults with celiac disease in the United States, where the disease is considered rare, and which is not well known.
Methods: We sought this information by distributing a survey. A questionnaire was distributed by way of a celiac newsletter, directly to celiac support groups, and through the Internet.
Results: Respondents (1,612) were from all United States except one. Seventy-five percent (1,138) were biopsy proven. Women predominated (2.9:1). The majority of respondents were diagnosed in their fourth to sixth decades. Symptoms were present a mean of 11 years before diagnosis. Diarrhea was present in 85%. Diagnosis was considered prompt by only 52% and 31% had consulted two or more gastroenterologists. Improved quality of life after diagnosis was reported by 77%. Those diagnosed at age > 60 years also reported improved quality of life. Five respondents had small intestinal malignancies (carcinoma 2, lymphoma 3) accounting for a relative risk of 300 (60-876) for the development of lymphoma and 67 (7-240) for adenocarcinoma.
Conclusions: Patients with celiac disease in the United States have a long duration of symptoms and consider their diagnosis delayed. Improved quality of life after diagnosis is common. An increased risk of developing small intestine malignancies is present.

Language: English
Publication Type: Journal article, research
Keywords: Celiac disease / Diagnosis / Quality of life / Survey

Kleveland PM
Children and adolescents with ulcerative colitis and Crohn disease
[Barn og ungdom med ulceros kolitt og Crohns sykdom]

Tidsskrift for den Norske Laegeforening 1998; 118(14) : 2160-2162
NIVEL: C 8184

This article addresses the problem that there is often a lack of awareness among people in general, parents and general practitioners about the possibility that a child suffers ulcerative colitis or Crohn's disease, because these diseases are relatively rare at a young age. There is therefore often an unnecessarily long delay before the young patient is seen by a general physician and then referred to a specialist centre where the diseases can be properly diagnosed, evaluated and treated. Chronic gastrointestinal symptoms caused by inflammatory bowel disease at an age where physical and mental growth are at a peak could result in serious negative effects not only on young patients' physical and mental development, but also on their self-esteem. The diseases adversely affect the quality of life of many young patients and their families and are a source of anxiety, emotional, behavioural, and psychological morbidity. Patients' organisations can help by providing additional information and support through special groups for young patients with inflammatory bowel disease.

Language: Norwegian
Publication Type: Journal article, general information and opinions
Keywords: Crohn disease / Ulcerative colitis / Children / Diagnosis / Quality of life / Psycho social problems

Rikken F, Loenhout JWA van
The treatment of Gaucher disease in The Netherlands
[Rapport ‘de behandeling van de ziekte van Gaucher in Nederland’]
Amstelveen: College voor Zorgverzekeringen, 2000
NIVEL: R 9400(40

This report describes a project about the treatment of Gaucher disease and the development of a clinical practice guideline. Of special interest are the summary and Chapter 7, in which a study is described about the quality of life of patients who suffer this disease.
Objective: This study was aimed at getting insight into health complaints and psycho social aspects.
Methods: Questionnaires were filled out at 4 different moments, before therapy (an enzyme replacement treatment) and 6 months, 12 months and 4 years after treatment. At the start 25 patients joined this study.
Results and conclusions: Many patients reported complaints like pain in bones and joints, fatigue and shortage of breath, and restrictions in activities of daily living. These complaints did not change over time. More than half of

the patients reported having a job, but all of them experienced restrictions. Problems in daily life activities did not change over time either. In conclusion, no much change was found after enzyme replacement treatment but improvement in quality of life was found after a few years.

Language: Dutch
Publication Type: Report
Keywords: Gaucher disease / Drug therapy / Quality of life / Questionnaire study

2.3.3 Psychosocial functioning and coping

Böcker FM, Seibold I
Assessment of "coping with disease" in patients with amyotrophic lateral sclerosis (ALS): on the use of an interviewer assessment rating scale
[Einschätzung der "Krankheitsbewältigung" bei Patienten mit myatrophischer Lateralsklerose (ALS): zur Anwendbarkeit einer Fremdbeurteilungsskala]
Psychotherapie, Psychosomatik, Medizinische Psychologie 1990; 40(8): 285-292
NIVEL: C 8186

Objective: To address the question how patients get along with a threatening condition like myatrophic (or amyothrophic) lateral sclerosis (ALS), a rare chronic progressive disease of the nervous system with a gradual loss of motor neurons, resulting in muscular atrophy, weakness and spasticity. Since the etiology is unknown and no curative treatment available, most patients die from respiratory failure within a few years.
Methods: After semi-standardised clinical interviews with patients and close relatives, two investigators gave independent descriptions of reported or observed "coping strategies" (rated as "not at all-barely possibly-probably-very probably present"). Considering patients assessed by both raters (N=17), significant agreement (Kendall's W) was achieved in three patients only.
Results: A comparison of aggregated answers (Chi^2-test) revealed different response sets, since one rater tended to choose extreme scale points, while the other preferred undecided answers. Differences of mean scores were observed in 8 out of 21 items, while significant correlations between investigators were obtained in another 8, of which only 2 items were rated as "probably present" in 50% or more of the patient sample. Two separately performed exploratory factor analyses (resulting in five factors each) made it obvious that both used different frames of reference for their evaluations. An

attempt to corroborate the validity of one of the two judgments based on external criteria failed too: Answers of both correlated significantly with measures of depression and anxiety (patients' self rating and relatives' assessment).
Conclusions: The data indicate response sets of the participating investigators rather than descriptions of coping behaviour of the patients examined. This unexpected result points to the well known fact that verifications of the inter-rater reliability of rating scales of similar design are necessary to avoid the interpretation of uncertain and irrelevant findings.

Language: German
Publication Type: Journal article, research
Keywords: Amyotrophic lateral sclerosis / Coping / Rating scales / Psychometrics

Cobb AK, Hamera E
Illness experience in a chronic disease: ALS
Social Science & Medicine 1986; 23(7): 641-650

Objective: To get insight into subjective experiences, adjustment and effects of the illness on social relationships of patients with amyothrophic lateral sclerosis (ALS), a devastating neurological disease.
Methods: A case study method was used in a 1-year longitudinal study of two individuals with ALS (N=2). Participants were interviewed in their homes every 2 months to examine the effects of the illness on relationships with family, friends and the health care system. Changing ideas regarding causation, the use and evaluation of various therapies, use of illness role models, spiritual changes and symptom experience were also explored. Kleinman's concept of explanatory models guides the analysis of the data.
Results: Each individual had a different idea about the cause of the disease. Participant B adopted a cognitive approach in keeping with the biomedical model, while participant A identified the medical system itself as potentially responsible for speeding the progress of her disease. Symptoms recognised and noted by participant A were physical signs; participant B's symptom recognition was functional in nature, with less concern for the physical progress of a symptom than with how it limited the ability to fulfill the role functions. Social relationships underwent radical change for both participants.
Conclusions: While most of the current research on ALS is disease-oriented, searching for a cause and cure, it is argued that the explanatory models of lay persons are illness-oriented, in which illness is seen as the experience of the disease. There should be a greater emphasis on evaluation of therapies within this model.

Language: English
Publication Type: Journal article, research
Keywords: Amyothrophic lateral sclerosis / Sick role / Coping / Interpersonal relations / Explanatory models / Interview study / Case study

Cohen DJ, Bruun RD, Leckman JF (eds)
Tourette's syndrome and tic disorders: clinical understanding and treatment
New York: Wiley, 1988
Chapter 12: Cohen DJ, Ort SI, Leckman JF, Riddle MA, Hardin MT. Family functioning and Tourette's syndrome. p. 179-196.
NIVEL: C 8208
Chapter 13: Silver AA. Intrapsychic processes and adjustment in Tourette's syndrome. P. 197-206.
NIVEL: C 8209
Chapter 15: Hagin RA, Kugler J. School problems associated with Tourette's syndrome. p. 223-236.
NIVEL: C 8210
Chapter 17: Meyers A. Social issues of Tourette's syndrome. p. 257-264.
NIVEL: C 8211

In this book different issues concerning Tourette's syndrome are addressed. The above mentioned chapters address the psycho social perspective.
In Chapter 12, Family functioning and Tourette's syndrome, the challenges posed on families of a member with Tourette's syndrome and the problems which are experienced in their role as child, brother or sister, husband or wife, and parent are described.

In Chapter 13, Intrapsychic processes and adjustment in Tourette's syndrome, the reactions, attitudes and psychological defences of the Tourette's patient are addressed. It is stated that the social adjustment of the patient not only depends upon the environment but also on inner forces.
In Chapter 15, School problems associated with Tourette's syndrome, educational issues are described; the barriers to learning and ways for adapting instructional strategies.
In Chapter 17, Social issues of Tourette's syndrome, the social issues that can have a major impact are described. These are divided into 2 categories: attitudinal and behavioural barriers, and community services to which patients can be referred, like special education or vocational rehabilitation.

Language: English
Document: Book Chapters

Keywords: Tourette's syndrome / Family / Attitude / Coping / Social adjustment / Education

Crandall CS, Moriarty D
Physical illness stigma and social rejection
British Journal of Social Psychology 1995; 34 (Pt 1): 67-83
NIVEL: C 8187

Objective: To discover which dimensions of stigmas are critical for causing social rejection using the stigma of illness as a model.
Methods: Students in psychology and human relations (N=415) were given a questionnaire packet, including 3 diseases vignettes from a total of 66. Subjects were asked to rate the illness on a variety of dimensions (e.g. contagious/not contagious, common/rare), and a measure of social rejection. In a second experiment, subjects responded to a case history of a fictitious disease that was either mild or severe, and was either behaviourally caused or not.
Results: Regression analysis revealed that two dimensions predicted rejection: the severity of the illness, and whether the illness was behaviourally caused (multiple R = .68). Results of the second experiment showed that behavioural causality and severity emerged as reliable, independent sources of rejection.
Conclusions: Diseases perceived to be severe or under personal control are most likely to lead to social rejection.

Language: English
Publication Type: Journal article, research
Keywords: Common diseases / Stereotyping / Prejudice / Sick role / Questionnaire study

Drotar D, Doershuk CF, Stern RC, Boat TF, Boyer W, Matthews L
Psycho social functioning of children with cystic fibrosis
Pediatrics 1981; 67(3): 338-343
NIVEL: C 8188

Objective: To describe the adjustment of children with cystic fibrosis as rated by parents and teachers compared to that of physically healthy siblings, normal children, and other chronically ill children.
Methods: Four groups were studied: cystic fibrosis patients (N=91), patients with other chronic illnesses (N=47), physically healthy siblings of cystic fibrosis patients (N=71) and physically healthy children (N=61). Questionnaires, consisting of items of behavioural adjustment, were sent to the parents of the children. Teachers filled in a School Behaviour Checklist.

Results: The findings indicate that children with cystic fibrosis achieved an age-adequate level of adjustment at home and school. Adjustment was largely unrelated to the severity of cystic fibrosis. As a group, chronically ill children had less adequate adjustment as rated by parents. However, severe adjustment problems were relatively rare.
Conclusions: These findings are consistent with a growing body of literature which suggests that children with cystic fibrosis can cope reasonably well with life tasks and that emotional disturbance is not an inevitable consequence of the disease. Future studies of the cystic fibrosis population should study factors which differentiate adjusted from maladjusted children.

Language: English
Publication Type: Journal article, research
Keywords: Cystic fibrosis / Children / Parents / Coping / Social adjustment / Interpersonal relations / Questionnaire study

Ek U
Emotional reactions in parents and children after diagnosis and treatment of a malignant tumour in the eye
Child: Care, Health and Development 2000; 26(5): 415-428
NIVEL: C 8189

Objective: To get insight into the emotional and psycho social effects on the parents of children with retinoblastoma, a rare malignant tumour in the retina usually presenting before 3 years of age, and on the children themselves.
Methods: This study includes all children diagnosed in Sweden between 1992 and 1994 (N=22). The parents and children were included in our longitudinal support programme immediately after the diagnosis was made. Parents were interviewed and children were observed by a psychologist about the parents' emotional reactions to the diagnosis and treatment and how the children reacted to the disease and treatment.
Results: Thirteen families were satisfied with the care offered, eight families expressed criticism. During the phase after the diagnosis the parents reported that they experienced psycho social problems; in fact, sixteen families reported symptoms of crisis. Developmental assessment appointments in the years following treatment gave opportunities to obtain guidance. All families except two welcomed the psycho social support which was offered. As far as the children's own reactions are concerned, most of them rapidly recovered after treatment. Between age 3 and 4 they gradually developed emotional reactions like anger and grief.
Conclusions: The parents' emotional reactions were strong and there were indications of crisis. The children's own reactions varied, but all of them

showed obvious psychological reactions to the loss of the eye. It is important to offer psycho social support to the parents to help them understand the disease and cope with it.

Language: English
Publication Type: Journal article, research
Keywords: Retinoblastoma / Children / Parents / Social adjustment / Emotions / Psychological stress / Interview study

Fahey S
The experience of women with primary biliary cirrhosis: a literature review
Journal of Advanced Nursing 1999; 30(2): 506-612

This article reviews the literature on the illness experiences of women suffering primary biliary cirrhosis (PBC), a serious and life-threatening illness that mainly affects women. Epidemiological data on the prevalence of the illness are unclear. The experience of women with this chronic illness has not been explored within nursing research. A review of the literature concerning PBC therefore is based on general themes relating to chronic illness. A chronic illness has two meanings: the symbolic significance and the consequences for the individual. The symbolic significance of PBC can be related to symbolism relating to the liver in general and to the general assumption that liver disease is related to alcohol consumption. The consequences for the individual woman with PBC have been described as following a disease management trajectory. This may include appreciating the major symptoms of the illness. The main symptoms of PBC are fatigue and pruritus. These are both insidious and debilitating symptoms of unclear aetiology that can cause women with PBC problems when seeking an illness explanation. The symptoms may also interfere with the woman's body image and her caring role. It is suggested that the factors that relate to PBC may result in social isolation for women with the illness.

Language: English
Publication Type: Journal article, review
Keywords: Liver cirrhosis, primary biliary / Women / Psycho social problems / Stereotyping

Hakola HP
Psycho social reactions in the spouses of patients suffering polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy
Journal of Clinical Psychiatry 1990; 51(1): 21-24

NIVEL: C 8190
Objective: To get insight into the psycho social reactions by family members of patients with polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy (PLO-SL), a rare hereditary disease affecting both brain and bones.
Methods: The spouses of 21 Finnish PLO-SL patients were interviewed. At the yearly follow-up examinations 13 spouses were interviewed. For the remaining 8 spouses, information was provided by patients of family members.
Results: The reactions included rejection and divorce in 8 cases and rejection without divorce in 1 case. Jealousy, violence, and alcoholism were common in male spouses.
Conclusions: It is suggested that some of the spouses' negative psycho social reactions may be lessened if the family is informed early about the nature of the disorder.

Language: English.
Publication Type: Journal article, research
Keywords: Bone diseases / Dementia / Cerebral sclerosis / Family / Psycho social problems / Interview study

Heijmans M, Ridder D de
Structure and determinants of illness representations in chronic disease: a comparison of Addison's disease and chronic fatigue syndrome
Journal of Health Psychology 1998; 3(4): 523-537
NIVEL: C 7598

Objective: To get insight into the structure and determinants of illness representations of patients suffering chronic fatigue syndrome (CFS) or Addison's disease (AD).
Methods: A structured interview technique was used.
Results: Considerable differences were found between the group of CFS patients (N=98) and the group of AD patients (N=63) with regard to their ideas about the identity, time-line, control/cure, and consequences of their illness. Despite these differences, the pattern of correlations among these four dimensions of illness representation was found to be similar for the two groups. Moreover, the strength of the correlations points to the coherent nature of illness representations. The relations between the illness representations, personal variables, and disease-variables were also explored. Regression analyses showed the dimensions of illness representation to be explained rather well by personal and disease-related variables. Disease-related variables were the most important predictors for the dimensions of

identity and consequences; personal variables showed strong associations with time-line and control/cure.
Conclusions: Although significant differences were observed between the two groups, the patterns of inter-correlation among the various dimensions proved to be the same; those reporting more symptoms and judging their illness as more serious also reported more serious consequences, found it more difficult to adapt to their illness and believed less in possibilities for control or cure. A belief in a more chronic time-line was also related to a belief in less control and more serious consequences. Higher scores on the identity and consequences dimensions strongly correlated with lower feelings of control and a more chronic perception of the time-line of the illness. The symptoms of AD are generally handled rather well by medication. It is therefore possible that AD patients initially see their disease as acute, with few consequences, and only later, after repeated symptom flare-ups, acknowledge the chronicity en potential uncontrollability. The results not only showed disease-related factors, but also personality characteristics and demographics to play an important role in illness representation.

Language: English
Publication Type: Journal article, research
Keywords: Addison's disease / Chronic fatigue syndrome / Sick role / Coping / Interview study

Helder DI
Living with Huntington's disease
Leiden: Universiteit Leiden, 2002
NIVEL: B 5120

Objective: To investigate the illness perceptions and ways of coping of patients suffering Huntington disease and of their spouses, to describe these mechanisms in explaining quality of life, and to get insight into the relationship between illness perceptions and coping on the one hand and the severity of the disease on the other hand.
Methods: Patients and their spouses were interviewed (N=77 and N=90) with orally administered questionnaires.
Results and conclusions: There was a strong relationship between illness perceptions of patients, i.e. ideas about their identity and treatment possibilities, and a feeling of well being. The illness perceptions of spouses did not influence their feeling of well being. The most common used coping strategy of both patients and spouses turned out to be acceptation of having the disease. Having the disease has a negative influence on emotional

behaviour, activities of daily living, communication and paid work. The feeling of well being of spouses was not negatively influenced.

Language: English
Publication Type: Dissertation
Keywords: Huntington's disease / Spouses / Quality of life / Coping / Sick role / Informal care / Interview study / Questionnaire study

Helder DI, Kaptein AA, Van Kempen GM, Weinman J, Van Houwelingen JC, Roos RA
Living with Huntington's disease: illness perceptions, coping mechanisms, and spouses' quality of life
International Journal of Behavioural Medicine 2002; 9(1): 37-52

Objective: To get insight into the impact of Huntington's disease (HD) on the quality of life (QOL) of spouses, from a behavioural medicine or health psychology perspective. It was hypothesised that spouses' illness perceptions and coping mechanisms will contribute significantly to the prediction of their QOL.
Methods: Illness perceptions, coping mechanisms, and the QOL of spouses of patients with HD (N=90) were assessed by means of the Illness Perception Questionnaire, the COPE, and the Medical Outcome Study 36-item Short Form Health Survey, respectively.
Results: After controlling for demographic and illness-related variables, coping mechanisms explained a significant amount of variance of spouses' role functioning.
Conclusions: Given these results, more empirical and longitudinal research is justified on coping mechanisms and illness perceptions of spouses living with Huntington's disease.

Language: English
Publication Type: Journal article, research
Keywords: Huntington's disease / Spouses / Quality of life / Coping / Sick role / Informal care / Questionnaire study

Hodgkinson R, Lester H
Stresses and coping strategies of mothers living with a child with cystic fibrosis: implications for nursing professionals
Journal of Advanced Nursing 2002; 39(4): 377-383

Objective: To explore current stresses and coping strategies used by mothers and to identify roles and strategies that nursing professionals could extend or adopt to support them and families of children with Cystic Fibrosis.
Methods: Semi-structured interviews were carried out with mothers of children with CF (N=17) who attended the regional cystic fibrosis hospital clinic. Interviews were analysed using the Framework method of analysis.
Results: The study suggests that major stresses for many mothers are feeling in the middle in terms of decision-making particularly concerning the genetic implications of CF, the burden of responsibility for parenting a child with a chronic illness, and coming to terms with a personal change in identity. The most commonly used coping strategy was seeking support from others including nursing professionals. Relationships with health professionals in secondary care were generally positive, with nurses often the first people mothers turned to when they had concerns and also key in interpreting medical information. Relationships with primary care health professionals appeared to be more limited with a role reversal described by some mothers who felt it their responsibility to educate and inform primary care professionals about CF.
Conclusions: This study suggests that nursing professionals in primary and secondary care need to look beyond the care of the child to the needs of the mother. It provides evidence to suggest that nursing professionals in primary and secondary care have a number of important roles to play as holders of hope, bridge builders and in providing continuity of care for such families. It also suggests strategies to strengthen the nurse-patient relationship.

Language: English
Publication Type: Journal article, research
Keywords: Cystic fibrosis / Parents / Psycho social problems / Coping / Social support / Nursing / Professional-patient relations / Interview study

Ievers CE, Drotar D
Family and parental functioning in cystic fibrosis
Journal of Developmental and Behavioural Pediatrics 1996; 17(1): 48-55
NIVEL: C 6445

This article reviews 31 articles describing studies on functioning of families and/or parents of children with cystic fibrosis (CF), compared to families of healthy children, pinpointing factors that predict quality of adjustment, or evaluating interventions designed to improve functioning. Commonly cited concerns included the difficulty of the treatment regimen, the terminal nature of CF, and the disruption of intra familial relationships. Parents of children with CF experienced greater stress and burdens than parents of healthy

children, yet parenting behaviour and family functioning were quite similar in CF and healthy control groups. Higher levels of distress, an avoiding coping style, and low levels of family support were associated with poor psychological adjustment. Recommendations for future research include: using multiple informants; using detailed, illness-specific measures and conceptual frameworks; and conducting studies of individual variation and interventions. Practitioners might consider performing periodic updates of how the family is managing as a whole.

Language: English
Publication Type: Journal article, review
Keywords: Cystic fibrosis / Children / Family / Parent-child relations / Psycho social problems / Coping / Social support

Jones P
The risk of suicide in people with haemophilia who are HIV infected
Aids Care 1995; 7 suppl 2: S157-S161
NIVEL: C 8191

This article addresses risk factors for suicide among HIV infected haemophilia patients, like easier access to analgesic drugs, loneliness, isolation and unemployment. Next, possible protective factors are outlined, which should be paid attention in haemophilia comprehensive care in order to reduce some of the factors associated with suicide risk. Protective factors include family ties, continuity of care, ease of specialist referral and reduction in uncertainty.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Haemophilia / HIV / Suicide / Risk factors / Continuity of care

Kiely L, Sterne R, Witkop CJ Jr.
Psycho social factors in low-incidence genetic disease: the case of osteogenesis imperfecta
Social Work in Health Care 1976; 1(4): 409-420
NIVEL: C 8192

Objective: To get insight into the psycho social implications of Osteogenesis imperfecta (0I), a rare hereditary disorder.

Methods: A cross-sectional survey was performed. The sample consisted of 0I-affected adults (N=13) and families with an 0I-affected child (N=21) who were interviewed.
Results: Findings revealed numerous psycho social concerns and problems in addition to their complex and lifelong medical problems. Specific problem areas differed according to the severity of the disease, type, and mode of genetic inheritance.
Conclusions: The results indicate the need for local as well as national policy changes through legislation and extensions of existing services for low-incidence disease groups such as 0I-affected persons. Implications for social work practice interventions call for greater involvement with genetic diseases, more aggressive approaches in case identification and service coordination, and performing longer range monitoring functions than is usually the case.

Language: English
Publication Type: Journal article, research
Keywords: Osteogenesis imperfecta / Genetic diseases / Adults / Children / Parents / Interpersonal relations / Psycho social problems / Employment / Social work / Interview study

Livneh H, Antonak RF, Maron S
Progeria: medical aspects, psycho social perspectives, and intervention guidelines
Death Studies 1995; 19(5): 433-452
NIVEL: C 8193

This article discusses progeria, a rare genetic childhood disorder that invariably results in the individual's death during early adolescence. The article begins by describing the major medical aspects of progeria. This is followed by a discussion of the psycho social implications of the disorder with particular emphasis upon grief-triggered reactions following the death of a child with progeria. The article concludes with an overview of psycho social intervention guidelines for caregivers who work with families of dying children and adolescents.

Language: English
Publication Type: Journal article, review
Keywords: Progeria / Genetic diseases / Children / Adolescents / Family / Grief / Psycho social problems / Coping / Counseling

Opjordsmoen S, Waehre H, Aass N, Fossa SD
Sexuality in patients treated for penile cancer: patients' experience and doctors' judgement
British Journal of Urology 1994; 73(5): 554-560
NIVEL: C 8194

Objective: To evaluate sexuality after successful treatment of penile cancer.
Methods: Post-therapy sexuality was evaluated in men (N=30, median age 57 years; range 28-75) treated for cancer of the penis 80 months previously (median; range: 11-225 months). Treatment regimes were: local excision/laser beam treatment, definitive radio-therapy, partial penectomy, and total penectomy. Patients underwent a semi-structured interview and completed three self-administered questionnaires (psycho social adjustment to severe illness [PAIS], mental symptoms [GHQ], quality of life [EORTC QLQ C30]). A global score of overall sexual functioning was constructed consisting of sexual interest, sexual ability, sexual satisfaction, sexual identity, partner relationship and frequency of coitus.
Results: In ten of twelve patients treated by irradiation the sexual global score was not or only slightly reduced compared with two of nine patients after partial penectomy and one of five patients with local surgery/laser beam treatment. All four patients who had undergone total penectomy recorded a severely reduced sexual global score. Of the six single domains, sexual identity and partner relationship did not change with increasing age, whereas the other scores of sexual life deteriorated as the patient became older. In the patients treated by irradiation doctors evaluated the patients' post-treatment sexuality to be more impaired than that experienced by the patients.
Conclusions: Within the limitations due to the small number of patients studied, radiotherapy seems to be the treatment of choice in limited cancer of the penis if preservation of sexuality is a major therapeutic aim. Physicians counselling patients with this rare malignancy need more information about treatment-related problems of sexuality after different therapeutic modalities.

Language: English
Publication Type: Journal article, research
Keywords: Penile cancer / Sex / Sex disorders / Surgery / Radiotherapy / Quality of life / Interpersonal relations / Interview study / Questionnaire study

Parker M
Families caring for chronically ill children with tuberous sclerosis complex
Family & Community Health 1996; 19 (3): 73-84

NIVEL: C 8195

Objective: To document how families cope with the stresses of caring for a chronically ill child with tuberous sclerosis complex (TSC), a rare pediatric neurological disorder. Families caring for TSC children have some special needs as well as some needs which are common to all families caring for a chronically ill family member.
Methods: A pilot study was performed in which members of seven families were interviewed; twelve interviews were conducted (5 married couples and 2 married women).
Results: The results show that family members giving care to a child with TSC report difficulties providing care; lack of spontaneity in family life; feelings of guilt, ambiguity, and chronic grief; variable support from formal and informal sources; and a profound impact on all aspects of family life. Many families feel they face an endless stressful, unpredictable and potentially chaotic family life.
Conclusions: This research may present new insights for health care professionals. It is hoped that the discussion will raise their awareness about TSC and encourage them to offer more effective help to all families caring for chronically ill members.

Language: English
Publication Type: Journal article, research
Keywords: Tuberous sclerosis / Children / Family / Psycho social problems / Interpersonal relations / Coping / Interview study

Saviolo-Negrin N, Cristante F, Zanon E, Canclini M, Stocco D, Girolami A
Psychological aspects and coping of parents with a haemophilic child: a quantitative approach
Haemophilia 1999; 5(1): 63-68
NIVEL: C 8196

Objective: To assess resources, stress, and coping in parents with a haemophilic child. Although haemophilia is a relatively rare hereditary disease, and is curable by blood products, the psychological and social problems of haemophilic patients and of their families are always serious. Anxiety for the risk of bleeding and the fear for infections transmitted by blood products cause stress and difficulty in coping with the situation.
Methods: In order to measure the dimensions related to family stress and resources, social desirability, tendency to depression, and anxiety, the subjects were administered the short-form of the Questionnaire on Resources

and Stress by Friedrich et al., revised by Saviolo & Cristante, along with the Social Desirability Scale, the Beck Depression Inventory, and the Ipat Anxiety Scale. The subjects of this study were 20 couples who were the parents of haemophilic sons. We analysed how the parents perceived the behavioural characteristics of their child, the differences between mothers and fathers in the questionnaire scores, and the correlation among the considered dimensions.
Results: For the mothers a negative relation between the subscales of the QRS and Social Desirability scale were observed; in addition, the dimensions of anxiety showed negative correlations with the Social Desirability scale. The correlations between the depression tendency and the QRS scales and between depression tendency and the anxiety scales were positive. Of particular interest were the correlations between the 'unconscious' anxiety scale and the QRS dimensions. For the fathers, there were fewer significant correlations and in general they were lower than those of the mothers. There was a positive correlation between anxiety and pessimism and between 'expressed' anxiety and depression tendency, while there was no negative relation between the subscales of the QRS and Social Desirability scale. The more the fathers are stressed the less they accept with consciousness and a sense of reality their child's disease.
Conclusions: The results show that the parents paid great attention to the problems of their sons, but mothers were more depressed and anxious about the disease of their child.

Language: English
Publication Type: Journal article, research
Keywords: Haemophilia / Children / Parents / Coping / Psychological stress / Depression / Anxiety / Questionnaire study

Teijlingen ER van, Friend E, Kamal AD
Service use and needs of people with motor neuron disease and their carers in Scotland
Health and Social Care in the Community 2001; 9(6): 397-403
NIVEL: C 8197

Objective: To get insight into the current use of services and the perceived needs of patients with motor neuron disease.
Methods: A national study of patients with motor neuron disease (MND) and their carers was conducted in Scotland in late 1996. A questionnaire covering areas such as: use of medical, social and voluntary services; use of or waiting for specialised equipment; satisfaction with provided services; and standard demographic data was used in face-to-face interviews

conducted by the four Scottish Motor Neuron Disease Association care advisers. The care advisers also assessed the respondent's level of impairment, using a standard instrument: the Amyotrophic Lateral Sclerosis Severity Scale (ALSSS). The data was analysed using SPSS-PC.
Results: In total 153 people participated (response rate 92%), and as expected with this patient group the study recruited more men (57%) than women. Sixty per cent of the sample was severely disabled as measured on the ALSSS. Health services did not meet the needs of respondents in 19% of the cases and social services failed to do so according to 24% of respondents. Eighty per cent of patients with MND had an identified carer. Nearly four out of 10 of these carers had their sleep disturbed regularly, and nearly a quarter of them would have liked to have more help. In addition, differences were found in service provision between the East and West of Scotland, and consequently differences in respondent's perceptions about the extent to which needs were met.
Conclusions: As found in similar studies, the formal health and social care sectors in conjunction with voluntary organisations are only partially managing the trajectory of patients with a rare progressive degenerative disease.

Language: English
Publication Type: Journal article, research
Keywords: Motor neuron disease / Amyotrophic lateral sclerosis / Health services needs / Patient satisfaction / Delivery of health care / Family / Interview study / Questionnaire study

Walker MB, Hilbert GA, Rinehart J
Face to face with Sturge-Weber syndrome
Journal of the Society of Pediatric Nurses 1999; 4(2): 74-82
NIVEL: C 8198

In this article a case is described of one family living with a child with Sturge-Weber syndrome (SWS), a rare progressive congenital disease that has as its defining feature a port wine stain. Moreover, the implications for nursing are discussed and a short overview is given of the available knowledge on family problems. Social support is critical for families facing overwhelming care needs. Families also need anticipatory guidance about child rearing, developmental milestones, decision making, and coping strategies. Additionally, families may need help in mobilising professional and family resources and in effectively using available services.

Language: English
Publication Type: Journal article, general information and opinions

Keywords: Sturge-Weber syndrome / Hereditary diseases / Children / Parents / Coping / Nursing / Social support / Case report

Webster DC
Sex, lies and stereotypes: women and Interstitial Cystitis
The Journal of Sex Research 1996; 33(3): 197-203
NIVEL: C 8199

This article offers a description of Interstitial Cystitis (IC), a relatively rare and poorly understood bladder condition, affecting 10 times more women than men. Symptoms of IC include urinary frequency, urgency and supra pubic pain. Many persons also have urethral and vulvar pain. Misconceptions may contribute to patients' difficulties obtaining an accurate diagnosis and may diminish the likelihood that they will access appropriate care. Many common symptoms of IC, including dyspareunia, can seriously compromise the ability to maintain a satisfying sexual relationship. Effective management of the disease requires accurate information and possible related sexual problems. Patients and their partners need support from health care providers for making necessary modifications in the sexual relationship that can greatly contribute to an improved quality of life.

Language: English
Publication Type: Journal article, general information and opinions
Keywords: Interstitial cystitis / Psycho social problems / Diagnostic errors / Sexuality / Social support

2.3.4 Social aspects

Combes PF, Desclaux B, Malissard L, Pons A
Influence of the diagnosis of Hodgkin's disease on the quality of life of cured patients
[Poids du diagnostic de maladie de Hodgkin sur la qualité de la vie des sujets qui en sont guéris]
Bulletin du Cancer 1977; 64(3): 395-408
NIVEL: C 8200

Objective: To get insight into the familial and socio-professional reintegration of patients who have been treated for Hodgkin's disease.
Methods: Questionnaires were sent to patients who had been treated for Hodgkin's disease (N=142). The questionnaire contained items about marriage, fertility, pregnancy, resumption of work and social security.

Results: Ninety questionnaires were returned (69%). On a family point of view, after treatment, the rate of single people was higher than in general population. Even though one could observe marriages, and feminine sterility was rare, the number of children in married couple remained low. There was no change in rate of house moving. Most of the cured patients returned to work. This fact was related to duration of treatment, but variations depended essentially on familial situation, kind of work and quality of previous social insurance. For two groups of people risk of unemployment was higher: agricultural labourers were more often dismissed and contractual civil servants met with more difficulties finding an occupation. Definitive invalidity was rarely found. Finally cured patients appeared most of the time comparable with the healthy population. Still, they met with important difficulties obtaining a loan or contracting a life insurance. Premiums are frequently raised and policies restricted.
Conclusions: Difficulties in former Hodgkin's disease patients are not related to organic sequelae. Cured patients still remain suspicious for their social surroundings and employers. Former patients prefer to hide the pathologic episode.

Language: French
Publication Type: Journal article, research
Keywords: Hodgkin's disease / Quality of life / Work / Employment / Social security / Questionnaire study / Social implications

Lowton K
Parents and partners: lay carers' perceptions of their role in the treatment and care of adults with cystic fibrosis
Journal of Advanced Nursing 2002; 39(2): 174-181

Objective: To explore the perceptions and experiences of lay care-giving amongst parents and partners of adults with Cystic Fibrosis who were being treated at a specialist CF centre in England.
Methods: Relatives of adults with CF (N=31) were interviewed in their own homes using an interview topic guide. All interviews were audiotape recorded and transcribed verbatim. Analysis of data was assisted by ATLAS-ti, a software package for qualitative research.
Results: Two main themes surrounding lay carers' role in treatment and care were identified. Firstly, the notion of lay carers giving 'expert' care, both in hospital and at home was recognised. Parents' expertise was greater than that of partners until the patient required intensive hospital interventions, when partner expertise increased. Secondly, the degree of lay carers felt inclusion

in the hospital consultation appeared to depend on the nature of their relationship with the patient and the patients' health state.
Conclusions: Lay carers are routinely performing tasks for adults with CF that were once the remit of trained nurses. Families need higher levels of nursing and social support when certain treatments are used at home. Attention needs to be directed to how lay carers of adult patients can be included in hospital consultations.

Language; English
Publication Type: Journal article, research
Keywords: Cystic fibrosis / Parents / Partners / Informal care / Social support / Interview study

Storhaug K
Aspects of living conditions among groups of disabled children and their families in Norway: family situation, mothers' health, financial assistance
Social Science & Medicine 1983; 17(23): 1837-1845

Objective: To get insight into the effects of chronic conditions on the family situation, the relationship between a mother's self-reported health, the medical problem of the child and family resources, and determinants of receiving social insurance and welfare assistance.
Methods: A survey of disabled children in Norway aged 0-19, representing ten different disabling conditions, was carried out between January 1976 and December 1978 (N=875). Disabled children with several common and rare chronic diseases were included. Parents of the disabled children were interviewed, medical records studied and the children examined.
Results: Mother's age, level of education, presence of disabled siblings, spouse's education and profession as well as emergency situations related to the disabled child's condition, appeared to be factors influencing the mother's health and therefore inevitably the family's ability to cope with the situation. Social insurance seemed to have been granted in a rather haphazard way; only families of children suffering from haemophilia, mental retardation, spina bifida and cerebral palsy seemed to have received fairly adequate social insurance benefits. Families of children suffering from juvenile rheumatoid arthritis, asthma, congenital heart disease and epilepsy had received less social insurance assistance than those in the other groups. One-parent families had received more social insurance than others. Families with children who were totally dependent on their parents, who had several diagnoses or had spent much time in hospital, had also been granted more social insurance. Welfare benefits distributed by local authorities had mainly

been given to families who were also receiving social insurance benefits and to families of children with brain damage. Almost half of all families expressed needs for welfare benefits which had not been met.
Conclusions: There seemed to be an under consumption of both social insurance and welfare benefits, particularly among some diagnostic groups.

Language: English
Publication Type: Journal article, research
Keywords: Chronic diseases / Genetic diseases / Children / Parents / Family / Quality of life / Social insurance / Social welfare / Survey

Varekamp I, Smit C, Rosendaal FR, Brocker-Vriends A, Briet E, Dijck H van, Suurmeijer TP
Employment of individuals with haemophilia in the Netherlands
Social Science & Medicine 1989; 28(3): 261-270

Objective: A study was performed to determine whether improvements in the treatment of haemophilia over the past 20 years have influenced the prospects of these patients in the labour market.
Methods: Surveys on the medical and social situation of haemophiliacs in the Netherlands were carried out in 1972, 1978 and 1985. Most of the patients participated in these surveys (N=276, N=403, N=716).
Results and conclusions: Trends in employment do not show either an increase in the number of employed haemophiliacs or a decrease in the number administratively defined as disabled. However, considering the influence of the economic recession on the position of the chronically sick on the labour market and the rise in the number administratively defined as disabled in the Dutch population, haemophiliacs perform well. Sick leave has decreased considerably. Although the employment rate for the group of haemophiliacs is lower than that for the general male population, the level of employment in relation to educational achievements is high and most of the employed do not feel limited in their daily job activities by the haemophilia. Physical mobility is a main factor influencing the employment status but other factors, such as the type of occupation or former occupation and prejudice against people with haemophilia, have to be considered.

Language: English
Publication Type: Journal article, research
Keywords: Haemophilia / Employment / Work / Activities of daily living / Disability evaluation / Survey

Wilson L
Cost-of-illness of scleroderma: the case for rare diseases
Seminars in Arthritis and Rheumatism 1997; 27(2): 73-84
NIVEL: C 8201

Objective: To determine the societal costs of scleroderma (SSc), a rare chronic connective tissue disease that affects approximately 98,000 Americans. Lack of reliable national databases limit rare disease cost studies, and this study suggests methods of using multiple data sources to assess the costs of rare diseases.
Methods: Primary and secondary data sources were used to calculate direct and indirect costs of SSc, including discounted lifetime mortality and morbidity costs. A prevalence-based, human capital approach was used in which a prevalence of 375 per million population was used as the base case for this study. Sensitivity analyses were used to vary parameters that are uncertain, such as prevalence, mortality, and labour costs.
Results: Annual direct and indirect costs of SSc in the United States were $1.5 billion. Morbidity represented the major burden, with $819 million (56%) of total costs. The current value of lifetime earnings lost was $179 million (12%) or $300,000 per death. Direct costs were $462 million (32%) or $4,731 per person annually, indicating that costs were spread over the long disease duration.
Conclusions: This study provides one model for the assessment of rare disease costs. Triangulation of data sources and sensitivity analyses are important for determining the costs of rare diseases. The high cost of SSc, despite its low prevalence, suggests that the burden of rare chronic diseases can be high. The high morbidity costs reflect the young age of onset of the disease as well as the need for treatments to decrease morbidity costs. Local shared databases and national surveys are needed to improve cost estimates of rare diseases.

Language: English
Publication Type: Journal article, research
Keywords: Scleroderma / Cost of illness / Cost-benefit analysis / Morbidity / Mortality / Employment / Sick leave / Cost model

APPENDIX 1 Keywords

All keywords were carefully chosen and tried out before the actual search strategies were performed.

Disease terms:
rare disease(s)
rare illness(es)
rare disorder(s)
rare progressive degenerative disease
congenital, hereditary, neonatal diseases and abnormalities

low prevalence or low incidence:
hereditary disease(s)
hereditary disorder(s)
hereditary illness(es)
chronic disease(s)
chronic illness(es)
chronic disorder(s)
genetic disease(s)
genetic illness(es)
genetic disorder(s)
hereditary disease(s)
hereditary illness(es)
hereditary disorder(s)

Health care structure:
health policy
orphan drug act
orphan drug production
orphan drugs

Health care process:
access to information
communication
communication barriers
consumer advocacy
consumer organisations
continuity of patient care
coordination of care
delivery of health care
disease management

health occupations/utilization
health services/utilization
health services needs and demand
information services
interpersonal relations
needs assessment
patient advocacy
patient care/psychology
patient care/trends
patient care/utilization
patient education
patient organisations
professional competence
professional role
social medicine
treatment possibilities

Health care outcome:
adaptation, psychological
attitude to health
client attitude
consumer attitude
consumer satisfaction
coping
diagnosis, differential
diagnostic errors
emotions
family relations
illness perception
life change events
patient attitude
patient-centred care
psychological
psychological functioning
psychology
psychosocial
quality assurance, health care
quality indicators, health care
quality of health care
quality of life
self-care
sick role

social adjustment
social environment
social functioning
social perception
social problems
social support
stress
treatment failure
wellbeing

Dutch keywords:
aandoeningen
aangeboren afwijkingen
chromosomale afwijkingen
chronische aandoeningen
chronische patiënten
erfelijke ziekten
niet gespecificeerde aandoeningen
somatische aandoeningen
weesgeneesmiddelen
weesziekten
zeldzaam/zeldzame
ziekte

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