

FOCUS
ON
RARE
DISEASES

The Dutch Steering Committee on Orphan Drugs

The Dutch Steering Committee on Orphan Drugs (Stuurgroep Weesgeneesmiddelen) has the mission to encourage the development of orphan drugs and to improve the situation of patients with a rare disease, in particular by improving the provision of information on rare diseases.

The term orphan drugs -in Dutch weesgeneesmiddelen- refers to medicinal products developed especially to diagnose, prevent or treat rare diseases.

The steering committee was established in April 2001 by the Ministry of Health, Welfare and Sport to nationally supplement the European Regulation on Orphan Medicinal Products. This multidisciplinary committee is today a significant network organization, putting the subject on the national agenda in various ways, serving as source of knowledge, and coordinating activities of parties involved in rare diseases. The Dutch website www.weesgeneesmiddelen.nl has a major role in these activities.

Simultaneously, the committee also participates in international exchange of knowledge and cooperation on treating rare diseases. And because these diseases are unaffected by borders, only a European and preferably global approach will lead to greater attention for and study of the ways in which patients with a rare disease should be treated.



Rare diseases mean *special* problems

Orphan drugs are medicinal products developed specially for rare diseases. These diseases by definition do not occur often, and are life-threatening and disabling.

Because of the seriousness and rarity of these diseases people who suffer from them (often young children) represent a special category of patients with special problems. These problems are special and serious to such extent that the Steering Committee on Orphan Drugs urgently draws attention to them and encourages parties to provide help and develop solutions.

This is really necessary. Although treatments have been developed for a number of sporadic metabolic disorders, heart and vascular diseases and cancer - partly as a result of the European legislation in 2000 - a remedy does not yet exist for most rare diseases.

What is rare?

In Europe a disease is labeled rare if no more than 5 in 10,000 citizens of the European Union suffer from it. This means that, taking all these diseases into consideration, some 30 million people in the European Union have to live on with such a - usually incurable, seriously disabling or hereditary - affliction. They are people who are entirely dependent on others' willingness not to 'overlook' them.

Rare diseases are usually *unknown*

Perhaps the biggest problem is the fact that most of these diseases are unknown to the general public and many researchers, doctors and care providers. Nobody is to blame because the number of disorders is estimated to be between 5,000 to 8,000. And because they present themselves so rarely medical practitioners may never have dealt with them in daily practice and thus be unfamiliar with them. Consequently, the symptoms of a patient suffering from a rare disease may not be recognized in time.

To avoid this situation, the steering committee publishes articles for doctors in several (mainly Dutch) magazines and contributes financially to several informative projects. Fortunately, ever more disorders are being described in a European database called Orpha.net.



A mother

She is the mother of a son of 19 years old. A son who is now severely physically and mentally handicapped because of a metabolic disorder called AGU.

'Between the age of three and four we noticed his development was slow. At that time we used to live and work in many different places abroad, so at first doctors believed that he was suffering from being 'dragged around' all the time.'

'Eighteen months later, after some infections and a urine analysis, all his cells proved to be missing a certain enzyme. Nothing was known about this. Rather coincidentally I ran into a scientific article describing 34 patients in Finland who suffered from the same disease. Today the Netherlands has 7 people suffering from this disorder, and no more than 250 patients worldwide.'

'You miss society's recognition and understanding. Years later I received some advice from a popular TV herbalist. She called me because she had seen a picture of my son. She said he was breathing wrongly.'

Rare diseases are often misunderstood

For patients, unfamiliarity with rare diseases often involves unpleasant consequences, which may interfere with the choice of the right treatment or dealing with the complaints.

Patients may spend a long time knowing nothing about the nature of their illness. Their direct environment (family and friends, acquaintances at school or at work) usually shows little sympathy for their complaints or difficulties.

In addition, unfamiliarity means symptoms are often not taken seriously or are ascribed to other possible diseases or causes. As a result patients may not be referred to the right specialists, or they may be submitted to a wrong treatment because of incorrect diagnosis. This is unfortunate because recognizing an illness on time and offering the right treatment may considerably reduce patients' suffering.

So the steering committee urges doctors to pay more attention to complaints, which cannot directly be ascribed to a certain illness. The sooner we know that a patient may be suffering from a rare disease the more efficient the approach will be.

The direct environment too should be more attentive to people having unusual health complaints.



A patient

In the mid-sixties his back started hurting. Medical tests showed nothing. So he went back and forth from the doctor, to the physiotherapist, from doctor to the chiropractor... This went on for fifteen years until he could bear it no more. An observant orthopedist, to whom he mentioned almost casually an intestinal hemorrhage he had had a few years ago, insisted on a blood test. The result: bad luck, a rare combination of Crohn's disease and the Bechterew disorder - the chances? 1 in 500,000. 'Both disorders are irreversible. Bechterew disables you and at the time there was hardly anything you can do about Crohn. It used to shorten your life considerably. Fortunately this is no longer the case. No information was known or exchanged about this disorder. Test results for instance were a mystery to the doctor and of course at first I did not have a clue of what was going on. My internist informed me that the Netherlands had a Crohn patients' association.'

Rare diseases often receive little attention

Rare diseases naturally present themselves only sporadically, and because little is known about many of them, they receive little attention, especially from researchers, politicians, the pharmaceutical industry, health insurers, doctors and care providers. Reasons include the fact that it concerns a small group of people who are easily overlooked. One may call it the impotence of the few. People are more likely to invest time, resources and ideas in problems that affect larger groups of people.

And all those who will know that developing a medicine usually takes ten to fifteen years and costs some 800 million euro on average will understand why the pharmaceutical industry is not keen on investing this amount of money in a solution which will only benefit a small group of patients.



A doctor

'This is also what makes it hard: in the case of rare diseases you cannot fully rely on what you have learned or experienced in practice. So a general practitioner needs something like a sixth sense, to be able to notice that something is not OK. To know that some inexplicable complaint might not be as innocent as it may seem. Some combinations of symptoms create such unusual effects that we need to wonder whether something truly exceptional might be going on. Recognizing those signals on time, taking them seriously and approaching them actively is indeed very important. It means talking to colleague doctors to find out whether they have had a similar case ... welcoming the information provided by patients themselves or by patients' associations ... and sending patients to specialists the moment you suspect something serious or abnormal is the matter or if a test shows a strange result.'

Promising developments are taking place

Does this mean that patients suffering from rare diseases are left to their own devices? Luckily they are not ...

Because there are always people who like to study what such diseases mean and what they do to patients.

The Internet is allowing patients from all over the world to contact each other, exchange information and experiences and support one other.

Because science continues to develop (e.g. gene technology and other biomedical developments), and because dedicated researchers always emerge to dive into a disease and eventually make a breakthrough.

Because quite some doctors are sufficiently attentive to the possibility of a rare disease thanks to practical experience, specialist literature and studies.

And because many parties (government, patients' organizations, primary healthcare, health insurers, the medical and pharmaceutical world) are taking initiatives to improve the position of people suffering from rare diseases.

Wherever possible, the steering committee seeks to encourage these developments.



The steering committee steers

Precisely with rare diseases, bringing together and mobilizing scattered knowledge and know-how is vital. Only this allows us to develop better methods to detect, monitor and treat these illnesses at an early stage.

With this in mind, the Dutch Steering Committee on Orphan Drugs is remarkably active in many fields.

So what does the steering committee do? In general, it takes on a coordinating role, gathers and exchanges information with patients' associations, pharmacists and doctors. It draws the attention of politicians, scientists and representatives of the pharmaceutical industry. The steering committee initiates research and organizes seminars, offering all parties access to the outcomes. It sits on international discussion and working groups and attends symposia.

Only an active approach and good cooperation with all parties involved across the world can protect patients with rare diseases from having to spend unnecessarily long times waiting for proper care.



The patients' organization on national level

The Netherlands is known for its high level of organization of patients. For instance it counts some 350 patients' associations, of which many deal with rare diseases.

The purpose of these associations is usually to collect as much information as possible about a certain disorder, bring fellow sufferers together, exchange experiences as to treatment methods and draw attention to patients' difficult situations.

In addition, many patients' organizations encourage scientific research and the development of an international network targeting a specific disorder. These disease-specific associations and umbrella organizations are again affiliated under a European joint venture, like Eurordis for instance for rare diseases.

A good example of a Dutch umbrella organization for rare diseases is the Dutch Genetic Alliance (VSOP).

What does the steering committee do at the national level?

The website www.weesgeneesmiddelen.nl covers most of the steering committee's activities in the Netherlands. The website includes a Dutch Orphan Drugs Database (Database Weesgeneesmiddelen) providing information on orphan drugs that are registered in Europe and information (if available) on relevant patients' associations, treatment centers or current research in the Netherlands. The steering committee works together with Orphanet Nederland contributing to the development of an extensive European online database.

Particularly fruitful are the so-called Orphan Topics and Orphan Cafes; these are multidisciplinary and sometimes open, informal get-togethers during which themes are scrutinized and discussed (e.g. costs or accessibility to orphan drugs).

Another example is the steering committee's contribution to a project which should lead on the one hand to the setup of a Dutch research program on orphan drugs and rare diseases, and on the other to the encouragement of researchers and (starting) companies in the Netherlands to develop orphan drugs.



The patients' organization on international level

Besides disease-specific associations, umbrella organizations (e.g. an association for all metabolic or neuromuscular disorders) and joint ventures between associations dealing with hereditary disorders or chronic illnesses also exist.

If a disorder is exceptionally rare, patients or parents sometimes call an international association into existence without establishing a national alliance. One such organization is the European Chromosome 11q Network.

What does the steering committee do at the international level?

The steering committee also works actively at the international level. Each year the committee is involved in the organization of an international workshop dedicated to serious (and especially rare) diseases. The workshop is held in different European capital cities under the flag of the European Platform for Patients Organizations, Science and Industry (EPPOSI).

The committee takes active part in the so-called E-Rare project; a project in which research funding organizations from eight different countries track research on rare diseases.

The steering committee also participates in several panel discussions and in international working groups such as the Coding Working Group of the Rare Disease Taskforce. And invests in cooperation and exchange of information with East-European organizations.

The committee encourages initiatives to found more multidisciplinary sister organizations in other countries.

Steering committee members and observers participate in working groups and committees of the European authority responsible for the evaluation of medicines, the EMEA, including the COMP (Committee Orphan Medicinal Products) and the WGIP (Working Group Interested Parties).

It is these contacts together with the other international contacts of individual members of the committee that allow the steering committee to carry out its activities to best effect at international level.



People with a mission

The Dutch Steering Committee on Orphan Drugs is expressly diverse as it deals with highly diverse diseases, which are similar in the sense that they all threaten to be underexposed.

The committee's composition is also diverse because all parties involved (from doctors to pharmacists, from patients' associations to health insurers and researchers) must contribute to help treating these diseases properly.

So committee members are from different disciplines; they are people with their own approaches and interests, (inter)national networks and fields of expertise. They have a high sense of involvement developed from daily practice.

Thanks to members' enthusiasm, knowledge, experience and contacts the steering committee is a unique forum capable of understanding almost the entire field of rare diseases.

Over the course of time this has allowed the Dutch Steering Committee on Orphan Drugs to develop into an authoritative institute in the wide field of rare diseases and orphan drugs. This means that the committee is the perfect discussion partner not only for the Dutch government, but also for other parties... at home and abroad.

People behind the steering committee

In this brochure (see inside cover) we briefly introduce individual members (and observers) of the steering committee.

If you wish to contact the Dutch Steering Committee on Orphan Drugs or if you have any questions on rare diseases or orphan drugs, please feel free to contact Jolanda Huizer or Sonja van Weely (secretariat) on working days.

Tel. + 31 (0) 70 349 5211

Or e-mail wgm@zonmw.nl

You will find a summary of the committee's main activities at www.orphandrugs.nl





Prof. H.G.M. Leufkens

Pharmaco-epidemiologist at the University of Utrecht. Chairman of the steering committee

'When it comes to enzyme research, the Netherlands can be proud of its record. We always had enthusiastic researchers and flag bearers but our culture does not have a strong tradition of benevolent patronage and we lack a national promotional fund that would provide further help.'



Dr. C.M.A. Rademaker

Pharmacist at the Wilhelmina Children's Hospital in Utrecht

'Many rare diseases present themselves at an early stage, which means childhood. Children and especially that small group of children suffering from rare diseases require tailored work when it comes to dosing and administering the right medicines. Tailored work usually requires innovation. Fortunately today there is also European legislation for children's medicines.'



Drs. R.H.M. Hendriks

Involved in health insurance and honorary president of the Association Internationale de la Mutualité

'Health insurers have to deal both with their legal tasks, and the equally unrelenting economic laws of the market system. I believe the answer to the question of how much a human life is worth cannot only be given by the market.'



H.J.J. Seeverens

Internist and policy adviser at the Ministry of Health, Welfare and Sport; Dutch member of COMPIEMEA (2000-May 2006)

'I am not a member of the steering committee, but an observer on behalf of the Dutch Ministry of Health. Within its remit the committee has managed to fulfill the significant function of the catalyst. Partly because of this more attention is now being paid to rare diseases and orphan drugs.'



Dr. J.P.T. Span

Clinical pharmacologist working at the Medicine Evaluation Board in The Hague and the University Medical Centre St. Radboud in Nijmegen

'I was involved in the registration of the very first orphan drug in Europe. Although orphan drugs should basically comply with the same requirements as ordinary medicines, the registration process today provides the possibility to give some medicines, which might never fully comply with the most strict scientific requirements, the benefit of the doubt.'



A. van der Zeijden

Council of the Chronically Ill and the Disabled in Utrecht; chairman of the International Alliance for Patients Organizations (IAPO)

'Being a patient myself I am familiar with the many obstacles patients might face. I consider it my task to keep the Dutch government fully aware to ensure it meets its commitments to European support policy.'



Prof. F.J.M. Gabreëls

Emeritus professor of pediatric neurology at the University Medical Center St. Radboud in Nijmegen

'The Netherlands has a good reputation when it comes to biomedical research but knowledge in this field is not being used optimally in practice. I am totally in favor of centers of excellence that assemble knowledge and experience in rare diseases. In this process a good referral policy is of paramount importance.'



Dr. M. van der Graaff

Manager of Innovation, Biotechnology and Genomics at Nefarma (Dutch umbrella organization of research oriented pharmaceutical industry) in The Hague

'Developments of molecular biology and information technology have created many breakthroughs. Think of the advances made in genetics. The level of knowledge is increasing and more rare diseases will be discovered. Unfortunately, the development process of orphan drugs is forced to lag behind, because it simply involves too much time and money.'



Ms. H. Meutgeert

Manager of the Association for Children and Adults with Metabolic Disorders (VKS) in Zwolle and working for the Dutch Genetic Alliance (VSOP) in Soestdijk; member of the Working Group with Interested Parties (WGIPIEMEA)

'Being the mother of a patient I sometimes wonder: isn't it odd that we know more about the moon than about how to repair certain damaged tissue in our bodies? Within the steering committee I focus primarily on the quality of healthcare. Patients and their families depend on this daily, for the rest of their lives.'



Prof. C.B.H.W. Lamers

Gastroenterologist at the Leiden University Medical Center

'Late diagnosis is a major problem. Take for instance the Zollinger-Ellison syndrome, a malignant tumor of the pancreas, which I discover about 15 times a year. The first visible symptoms are similar to those of a gastric ulcer. It can only be cured if action is taken on time.'



Drs. A.R. Schuurman, MBA

Sector Manager of the CVZ (Health Care Insurance Board) in Diemen; chairman of the medicine evaluation committee (MEDEV) of the European Social Insurance Fund

'In the Netherlands people suffering from a rare disease are also entitled to high-quality insurance and reimbursement. I hope to be able to properly realize this through the Dutch Steering Committee on Orphan Drugs.'



Ir. J.G. Hanstede

Chair of BioFarmind (the Dutch Association for the Biopharmaceutical Industry) in The Hague

'Biofarmind is an interest group and network organization for the biotechnological and pharmaceutical industry. Developments in the field of molecular diagnostics are taking place at high speed. Understanding man's genetic profile allows us to better refine the diagnosis and so better tune our solutions.'



Dr. A.C.G. Voordouw

Board for evaluation of medicines; Dutch member of COMPIEMEA from May 2006

'The Committee for Orphan Medicinal Products, on which I sit as a Dutch delegate, seeks to encourage the development of orphan drugs in Europe. As a member of COMP and observer on the steering committee, I hope to be a link between what is being done at national and international levels.'



Drs. J.S. Huizer

Secretary of the Dutch Steering Committee on Orphan Drugs; member Coding Working Group (Taskforce Rare Diseases)

'In the late nineties my work at the patients' organization VSOP got me involved in rare diseases. At the time treatments hardly existed, but now several orphan drugs have been developed across Europe. New bottlenecks presented themselves such as availability and reimbursement. The task of the steering committee is to signal matters and mediate between parties.'



Dr. S. van Weely

Secretary of the Dutch Steering Committee on Orphan Drugs; member organizing committee annual workshops of EPPOSI

'As a biochemist I spent years studying a rare disease that could not be treated 15 years ago; today several treatments have been developed. This experience encouraged me to improve the situation of people who suffer from rare diseases. By working together with national and international organizations and exchanging information on research, healthcare and treatments.'

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