ZOOM

STANDING UP FOR PEOPLE WITH RARE DISEASES

King Baudouin Foundation
Working together for a better society

WHAT MEANS 'RARE'?

A disease is rare when it affects less than 1 in 2,000 people in the population. Nevertheless, people with a rare disease are far from rare. Quite the opposite. There are an estimated 6000 to 8000 rare diseases, so 6% to 8% of the world's population suffer from one 'orphan disease' or another. In the case of Belgium that adds up to more than half a million people.

There is still a lot that remains unknown about rare diseases. Due to a lack of knowledge, a correct (and timely) diagnosis is often not made. In many cases there is no suitable treatment at all. Or, if it does exist, patients do not manage to access it. What is more, relatively little scientific research is carried out into rare diseases.

AT A GLANCE

The King Baudouin Foundation has been working in this area in various ways for many years, providing robust support for research into rare diseases and patient care.

- The **Fund for Rare Diseases and Orphan Drugs** brings together all those involved in the area of rare diseases and has laid the foundations for the Belgian Plan for Rare Diseases.
- To provide strong support for **research**, the **Generet Fund** awards an annual prize of 500,000 to 1 million euros for pioneering research in the area of rare diseases at a Belgian university. Several different Funds have also been set up to promote research into specific rare diseases.
- Thanks to the pilot project **Mind the Gap!** a list of priority research questions which are widely considered to be important has been produced for the rare genetic disorder Tuberous Sclerosis Complex (TSC).

DID YOU KNOW...

44%
HAVE RECEIVED AN INCORRECT DIAGNOSIS
OR ALMOST **1 IN 2 PATIENTS**
WITH A RARE DISEASE.

1 IN 3
RECEIVED INCORRECT TREATMENT.

22%
SAW AT LEAST 5 DOCTORS BEFORE
THE DIAGNOSIS WAS MADE.

7%
SAW 10 DOCTORS OR MORE.

36%
HAVE HAD TO STOP WORK OR DO LESS WORK.
IN **1 IN 4 CASES**
A FAMILY MEMBER STOPPED WORK TO CARE FOR
A PATIENT WITH A RARE DISEASE.

WHERE ARE WE TODAY?

BELGIAN PLAN FOR RARE DISEASES

Based on the proposals set out by the Fund for Rare Diseases and Orphan Drugs, Belgium launched an action plan for rare diseases in 2014.

The Plan consists of 20 actions in various areas, such as improving access to diagnosis and information; optimising care by sharing and concentrating expertise; improving the management of knowledge and information about rare diseases through development of a central register for rare diseases etc.

Slowly but surely, this Plan is leading to concrete achievements:

- People suffering from cystic fibrosis, rare neuromuscular disorders, metabolic diseases, haemophilia, kidney diseases in children, refractory epilepsy or spina bifida can now be managed by *referral centres or rehabilitation institutions with an NIHDI agreement*.

- In 2016 seven University Hospitals received recognition for a '*Rare Diseases Function*'. The Rare Diseases Function is an access point for patients with an undiagnosed disease: it aims to make a diagnosis and develop a treatment plan, or else to make a referral to a specialist with proven expertise if available.

- In late 2017 the first five '*Rare Diseases Networks*' were started up. The Networks bring together the expertise of all the organisations involved in reception, guidance and treatment of patients with a rare disease. The Networks are organised according to groups of rare diseases. A similar initiative has also been initiated in the Wallonia-Brussels Federation.

- The European Union has developed '*European Reference Networks*' (ERNs). The ERNs bring together the strengths of centres of expertise at the European level by sharing their specialist knowledge and resources in the area of rare diseases.

Eva Schoeters,
President of RaDiOrg:

"The changes taking place as the Belgian plan for Rare Diseases and Orphan Drugs is implemented can only be felt to a limited extent by individual patients, who are of course living with a strong 'sense of urgency'. Thanks to the plan, however, more attention is being devoted to rare diseases. That gives us hope!"

PROMOTING RESEARCH INTO RARE DISEASES

Generet Fund - prestigious annual prize for pioneering research into rare diseases

The Generet Fund is putting Belgian research into rare diseases on the international map. Each year it awards a Prize of 500,000 euros to a top researcher. This amount can also be doubled after an interim progress report is approved.

The Fund does not determine which condition has to be addressed by the research. All (groups of) rare diseases are eligible.

The Generet Prize was for the first time in 2018 awarded to prof. Miikka Vikkula of the Laboratory of Human Molecular Genetics, de Duve Institute (UCL).

For more than twenty years, he has been conducting research on vascular anomalies (angiomas), which are chronic and rarely curable diseases. These anomalies can affect any tissue or organ, and have a major negative impact on patient's quality of life.

Professor Vikkula aims to identify the causes of these diseases to provide input for the development of new therapies. As the patients and their quality of life are at the core of his research, he was one of the founders of VASCAPA, an association for patients with a vascular anomaly.

Funds focusing on a specific rare disease

In recent years several Funds have been set up within the King Baudouin Foundation to support research into specific rare diseases. The individuals setting up these funds are often people who have experience of a rare disease themselves or among those close to them.

Although they have no guarantee that they will benefit from the results of the research themselves, that does not stop them from getting involved for the benefit of all patients. The focus and working methods differ from one Fund to another and these are based on the wishes of the founder and the key needs of patients.

Here are some examples:

The **Fund Alphonse & Jean Forton** for research into cystic fibrosis has supported no less than 53 research projects since it was set up in 1997, for a total of 8 million euros. This Fund can be described without exaggeration as having given a significant boost to research into cystic fibrosis in Belgium: the Fund has given rise to a very active research community with an international reputation. Partly as a result, Belgium has become a major centre for research into cystic fibrosis.

Stefan Joris,
Belgian Cystic
Fibrosis Association:
"The Fund J&A Forton has played an essential part in improving life expectancy and quality of life for patients. By joining forces with the Belgian CF Association, the call for projects targeting scientific research into cystic fibrosis becomes even more significant at the European level."

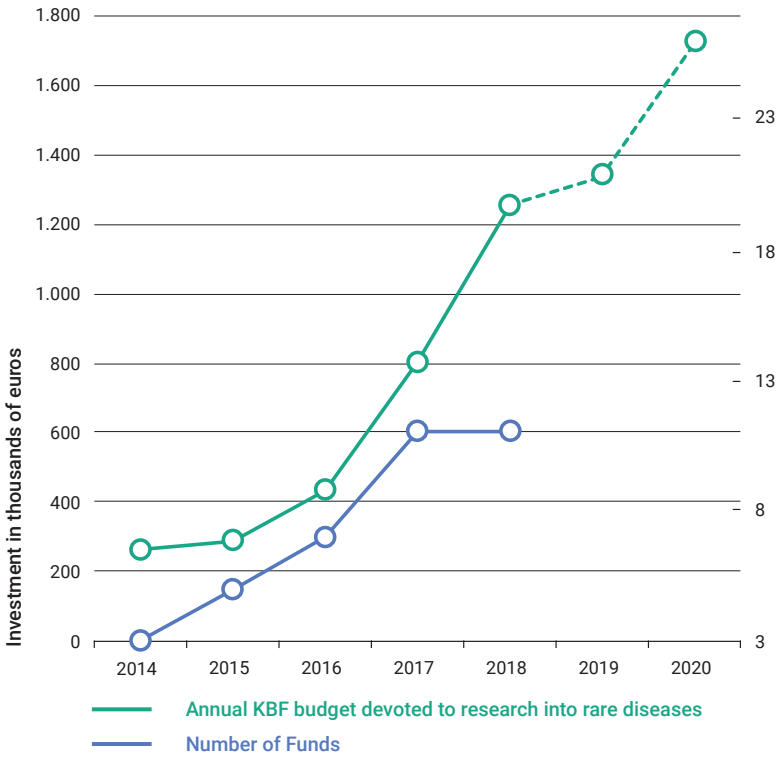
The **Fund Vlinderkindje** has managed to persuade international expert Dennis Roop (University of Colorado - US) to initiate research into the hereditary skin disease Epidermolysis Bullosa simplex (EBS). Dennis Roop is looking for a curative treatment for this condition, through stem cell therapy.

Sylvie De Maegd,
founder of the
Fund Vlinderkindje:
"Through the Fund we want to support scientific research and at the same time increase awareness in society. EBS is a very visible disease. People who are not familiar with it immediately think that the disease is infectious."

The **Fund 101 Genomes** was founded by the parents of little Aurélien, who was born with Marfan syndrome. The Fund aims to make a bio-informatics platform available to all interested researchers. The platform consists of a database containing genomic data and medical data from patients with this disease - the target is to gather data from at least 101 patients.

Romain Alderweireldt,
founder of the
Fund 101 Genomes:
"Without good fundamental data there can be no good research. With this bioinformatics tool researchers do not need to build their own cohort of patients and carry out the DNA- sequencing analysis themselves."

Philanthropy is also making progress in the area of rare diseases. This is clear, for example, from the increased number of Funds for rare diseases at the King Baudouin Foundation and the annual increase in the budget available for investment in research into rare diseases.



PROSPECTS FOR THE FUTURE

Determining research needs through dialogue

Through the 'Mind the Gap!' process the King Baudouin Foundation has tested out an innovative method of setting research priorities. One of the pilot projects focused on the rare genetic disorder Tuberous Sclerosis Complex (TSC).

All those involved, both patients and their family members, researchers, doctors and care providers, were involved in the dialogue and considered how research can respond better to the real needs of patients, those close to them and also care providers. This resulted in a list of 15 research questions for TSC which everyone views as a priority.

Right at the top of the list was the demand for research into the behavioural, learning and neuropsychiatric problems affecting TSC patients.

Together with the Fund Doctor & Mrs Charles Tournay-Dubuisson, the King Baudouin Foundation has decided to make 600,000 euros available over a period of four years for research in this area.

Prof. Anna Jansen, coordinator of a multidisciplinary TSC consultation at UZ Brussels: "In view of the many areas of overlap between different rare diseases, this model exercise for TSC can also provide added value for other conditions. The pilot project has created an impetus for more dialogue between those affected and professionals."

Research brings hope

Research into rare diseases is necessary, because the needs in this area are still considerable. Certainly in comparison with more common conditions, research into rare diseases is still not very popular and research budgets are too limited.

Nevertheless we have reason for hope. With protein therapy, immune-modulatory therapy, gene therapy, cell therapy etc. new, innovative therapeutic opportunities are opening up. These give patients prospects for the future, even though that future sometimes seems to be a long way off.

A lot of work is also being done on care and the organisation of care, to identify and remedy a number of gaps that exist.

READ MORE

- **Belgian Plan for Rare Diseases: to read at www.radiorg.be and www.health.belgium.be (in French and Dutch)**
- **Mind the Gap! – Working together to set research priorities for Tuberous Sclerosis Complex (TSC)**
- **Recommendations and Proposed Measures for a Belgian Plan for Rare Diseases – Final report**

You can download these two latest publications free of charge from **www.kbs-frb.be**

KING BAUDOUIN FOUNDATION

The King Baudouin Foundation's mission is to contribute to a better society.

The Foundation is an actor for change and innovation, serving the public interest and increasing social cohesion in Belgium and Europe. We seek to maximize our impact by strengthening the capacity of organizations and individuals. We also stimulate effective philanthropy by individuals and corporations.

The King Baudouin Foundation is a public benefit foundation. The Foundation was set up in 1976 on the occasion of the 25th anniversary of King Baudouin's reign.

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