RARE DISEASES Magazine

EDITION 2026

The new Belgian Rare Diseases Plan: an integrated vision for care and policy

Multidisciplinary care for epidermolysis bullosa

Blood spot screening: crucial early detection in newborns

UZ LEUVEN

Dear reader,

With this new edition of the UZ Leuven Rare Diseases Magazine, we would like to update you on recent policy developments and initiatives at our hospital. The magazine also serves as a reference resource and information source for referring clinicians.

We are pleased that a new Belgian Rare Diseases Plan was published at the end of February. It sets a course towards faster diagnosis, stronger multidisciplinary collaboration and better care coordination for all patients with a rare disease. Expertise by disease or disease group will become easier to locate through national mapping. This initiative starts this year for the recognised function hospitals and will provide important support for referring clinicians.

We are also making progress in data collection and registration. Over the past year, UZ Leuven has worked hard to improve the flow of data to the national rare diseases register. This register is an essential tool for better understanding the epidemiology of rare diseases in Belgium and, in turn, enabling data-driven policy decisions that genuinely respond to the needs of patients and healthcare professionals.

We are also proud that the KU Leuven Institute for Rare Diseases is now fully up and running. It brings together research, education and innovation at our university and reinforces UZ Leuven's role as an academic centre of expertise.

In this edition, you will also find practical information on referral and key figures about our activities. We hope the magazine will inspire and support you in your daily practice.

Happy reading!

Would you like to receive a printed and/or digital copy of the Rare Diseases Magazine? You can subscribe or unsubscribe via forms.office.com/e/uPDcUbvFhp.

Composition of the UZ Leuven Rare Diseases Bureau

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Chief Medical Officer - Biobank
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Institute for Rare Diseases

Previous editions

In previous editions of the Rare Diseases Magazine, you could discover, among other things:

- ✓ what a rare disease is and what the European, Belgian and Flemish stakeholder landscape looks like (edition 2024).
- ✓ the role played by the Centre for Human Genetics in the care and genetic diagnosis of rare diseases (edition 2024).
- ✓ how innovative DNA and RNA therapies are making a difference for rare diseases (edition 2025).

Did you miss the previous editions, or would you like to read them again? A digital version is always available at **uzleuven.be/en/rare-diseases**

RARE DISEASES AT UZ LEUVEN

300 DOCTORS IN VARIOUS DISCIPLINES

> 50 MULTIDISCIPLINARY TEAMS

33,000 PATIENTS WITH RARE DISEASES PER YEAR

IN 2024
12,931 NEW DIAGNOSES

10,079 ADULTS

2,852 CHILDREN

46 REFERENCE AND EXPERTISE CENTRES PUBLISHED ON ORPHANET

10 NIHDI CONVENTIONS FOR MULTIDISCIPLINARY CARE FOR RARE DISEASES

How can you refer a patient with (suspected) rare disease to UZ Leuven?

Without diagnosis of rare disease

1 If symptoms and clinical signs do not point to a single organ: refer to the general consultation in **general internal medicine (adults), paediatrics or genetics**.

2 If there is a clear clinical suspicion of an organ-specific disease: refer to the relevant **organ specialist**.

Registration via form

✓ If you suspect a rare disease but do not know where the patient can best be seen, use the **registration form for referring doctors**. Based on this, we will tell you more about the most appropriate consultation (at UZ Leuven or, if needed, elsewhere in Belgium or Europe).

Registration form for referring doctors via www.uzleuven.be/en/rare-diseases or directly at qr.uzleuven.be/bgGAs9

✓ Patients can also register themselves via the **patient registration form** (upload of a doctor's referral letter or relevant medical report required).

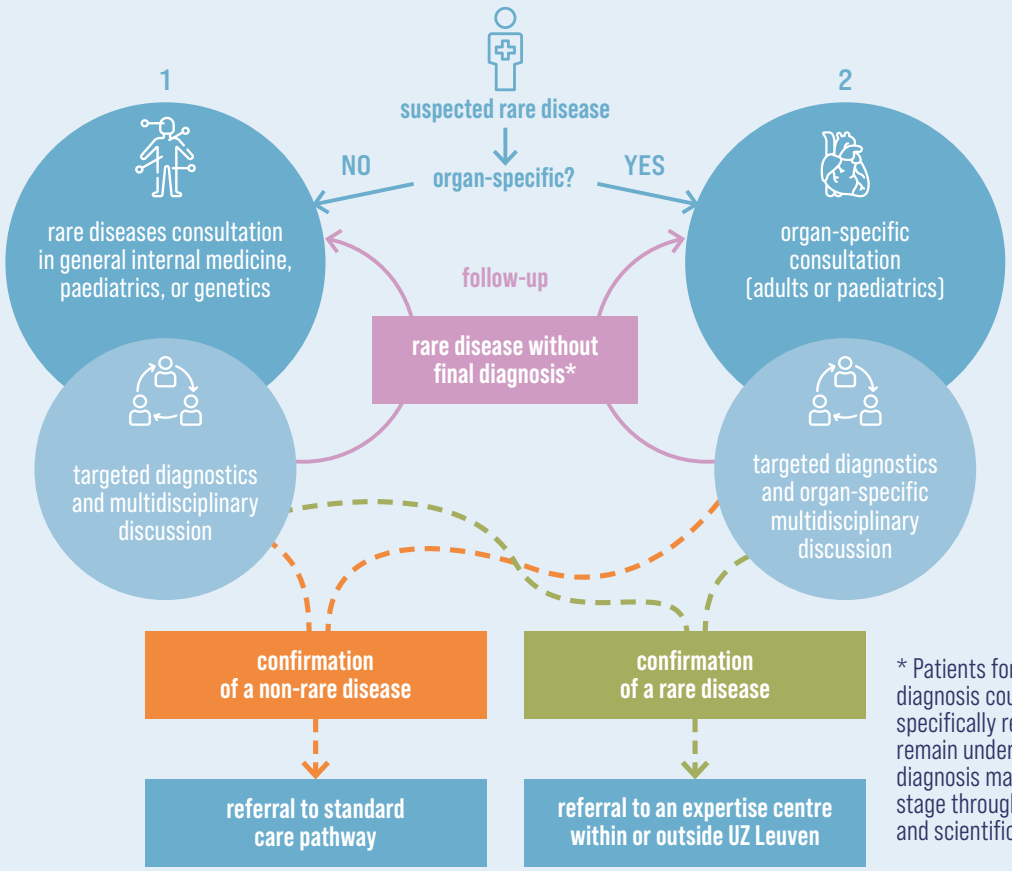
Patient registration form via www.uzleuven.be/en/rare-diseases or directly at qr.uzleuven.be/bgGBOj

With a rare disease diagnosis

✓ Where there is an established diagnosis, refer directly to the **team specialising in the condition**.

An overview of the teams can be found at www.uzleuven.be/nl/zeldzame-ziekten/met-diagnose (in Dutch)

✓ Some diseases are so rare that **no dedicated specialist consultation exists**. We may nonetheless have the relevant expertise in-house with an individual specialist. Please contact us at **rarediseases@uzleuven.be**.



* Patients for whom no final diagnosis could be made are specifically registered and remain under follow-up, so that a diagnosis may be made at a later stage through new technological and scientific developments.

The new Belgian Rare Diseases Plan: an integrated framework for clinical, policy and patient-centred progress

At the end of February, a renewed **Belgian Rare Diseases Plan** was published. For the period 2026-2030, it provides a strategic framework that affects clinical practice, health policy and the work of patient organisations alike. Rare diseases affect an estimated 500,000 people and therefore represent a substantial challenge for the healthcare system. The plan is structured around six domains whose initiatives, taken together, are meant to deliver a more coherent, accessible and inclusive approach.

1. **Faster and more accurate diagnosis**
   Focused on better recognition of rare diseases and more efficient referral (e.g. raising awareness among GPs, better access to diagnostics).
2. **Mapping and strengthening expertise**
   Building a national map of expertise so that healthcare professionals and patients know where specific knowledge is available.
3. **Better care coordination and multidisciplinary care pathways**
   Developing integrated care pathways, including care coordinators and clear points of contact.
4. **Access to innovative treatments**
   Faster assessment and wider availability of orphan medicines and new therapies.
5. **Better information, support and patient participation**
   Actively involving patients and their loved ones as experts by experience and improving access to information and support.
6. **Governance, monitoring and quality improvement**
   A structural policy framework with clear roles for the FPS, NIHDI, Sciensano and the rare diseases function hospitals, including follow-up of 25 concrete measures.

What does this mean for UZ Leuven?

For UZ Leuven, the plan means a further strengthening of the rare disease reference centres, which will grow into recognised **multidisciplinary centres of expertise**. These centres will develop clinical guidelines, support training and act as hubs for complex cases. The development of a national map of expertise and the financing of care coordination create new opportunities to consolidate internal structures and national and international collaborations.

What it means for referring clinicians

For referring clinicians, the plan offers a more transparent care landscape. The national cartography makes clear where specific expertise is located, allowing them to **refer patients more quickly and more accurately**. Digital decision-support tools, broader access to genetic and genomic tests, and the involvement of care coordinators improve the continuity and efficiency of complex care pathways.

A clearer view for policymakers

For policymakers, the plan provides a tool to reduce fragmentation in the care landscape. Further development of the **national rare diseases register** should improve epidemiological insight, enable quality monitoring and make policy planning more evidence-based. The plan aligns with European strategies and encourages participation in international networks such as the European Reference Networks (ERNs), strengthening Belgium's position within European health policy.

The patient as a full partner

For patient organisations, the plan explicitly recognises their role as structural partners. They are involved in setting priorities, evaluation and policy development. This approach reflects a broader move towards patient-centred care, in which lived experience is recognised as a fully fledged source of knowledge. In addition, psychosocial support is given a more prominent place, with attention to work, education, family and financial capacity.

Collaborating in research

The plan emphasises the importance of **translational research**, encouraging collaboration between academic centres, industry and clinical practice. This should lead to faster implementation of innovative diagnostics and therapies.

Overall, the plan offers an integrated policy framework that responds to the needs of healthcare professionals, policymakers and patients. Its effectiveness will depend on sustainable financing, intersectoral collaboration and a robust evaluation culture, but it represents an important step towards timely, high-quality and inclusive care.

The plan was developed through collaboration between:

- ✓ the FPS Public Health
- ✓ the NIHDI
- ✓ Sciensano
- ✓ the policy unit of the Minister for Social Affairs and Public Health
- ✓ the eight hospitals recognised as rare diseases function hospitals (including UZ Leuven)
- ✓ RaDiOrg, the Belgian umbrella association for patients with a rare disease
- ✓ the King Baudouin Foundation

Read the full plan here (in Dutch):
health.belgium.be/nl/nieuws/2026-2-plan-zeldzame-ziekten-2026-2030

Cartography of expertise in rare diseases: a compass for the care landscape

With the launch of the new Belgian Rare Diseases Plan, a crucial component of the policy is now taking concrete shape: the cartography of expertise. For the first time, this national exercise is meant to provide a complete, transparent and validated overview of where broad expertise exists in Belgium for specific rare diseases or clusters of conditions. This ambitious exercise is essential for better care coordination, policy planning and patient-centred information.

The methodology has been carefully developed by the rare diseases functions, the patient association RaDiOrg, the FPS Public Health, the NIHDI, Sciensano and the policy unit of the federal cabinet for Public Health.

What exactly does the cartography capture?

The focus is on hospitals recognised as a rare diseases “function” or “reference hospital”. A “centre” is defined as any multidisciplinary team within such a hospital that has *“broad expertise*

regarding a rare disease or a cluster of rare diseases". It is therefore not about expertise in a single treatment or a single age group, but about care across the full pathway.

To be included in the cartography, centres must demonstrate their expertise using an extensive set of **indicators**, including:

- ✓ patient and procedure volumes
- ✓ the presence of a multidisciplinary team
- ✓ care pathways (including patient-facing information on them)
- ✓ collaboration with other lines of care, hospitals and international networks
- ✓ participation in case discussions
- ✓ quality indicators
- ✓ scientific output, grants and participation in registers
- ✓ collaboration with patient organisations

Why is the cartography important?

For **doctors**, the cartography offers a clear overview of where expertise is located. This makes referral and collaboration easier. In addition, the indicators place the emphasis on multidisciplinary care, quality assurance and participation in scientific research.

For **policymakers**, the cartography provides an objective basis for future recognition, funding and network-building.

For **patient organisations**, the exercise creates visibility and clarity. The indicators explicitly require structural collaboration with patient associations, anchoring their contribution to quality improvement.

Outlook

The timetable provides for submissions to start this summer. These will be followed by analysis, evaluation, validation and publication of the results on Orphanet and on the websites of the FPS Public Health, the NIHDI and the rare diseases functions. According to the current plan, the cartography should be completed and visible to the public by June 2027.

In this way, the cartography becomes one of the foundations of the new Rare Diseases Plan: an instrument that makes expertise visible, exposes gaps and stimulates collaboration. It is a necessary step towards a more transparent, coherent and future-oriented care landscape.

Stronger together: multidisciplinary care for epidermolysis bullosa

Epidermolysis bullosa, or EB, is a rare inherited skin disease in which the skin is extremely fragile. UZ Leuven is the only recognised centre of expertise for EB in Belgium, with an official NIHDI convention. But what does that recognition mean in practice? We take a look behind the scenes, where a whole team of specialists is ready to support patients intensively, from birth right through to adulthood.

A whole team by your side

For a child born in Belgium with EB, the pathway quickly becomes clear: that child is referred to UZ Leuven. As the only recognised centre of expertise in our country, the hospital provides

Did you know...

- ... epidermolysis bullosa (EB) is a rare inherited condition in which even the slightest pressure or friction can cause painful blisters and wounds.
- ... EB has four subtypes: simplex, junctional, dystrophic and Kindler. The impact varies greatly: from limited blistering with a normal life expectancy to severe, life-threatening forms with complications such as feeding difficulties, skin tumours and organ failure.
- ... there is currently no cure or medicine that addresses all symptoms. Care therefore focuses entirely on support and relief.

specialised multidisciplinary care that is available nowhere else. Dr Caroline Colmant, dermatologist and coordinator of the NIHDI convention, explains their distinctive approach: “We always make the diagnosis together with the whole team. That is no empty promise; it is the very concept on which the EB team is built.”

“With a new patient, the parents immediately meet the whole team: dermatologist, nurse, neonatologist and geneticist”

Collaboration starts even before the diagnosis is confirmed. Newborns with suspected EB are admitted to the neonatal intensive care unit (NICU). There, neonatologist Prof. dr. Gunnar Naulaers coordinates the care process. Together with dermatologist Dr Colmant and clinical geneticist Prof. dr. Ellen Denayer, he explains to the parents what EB is, which type their child has, what that means for the future, and who their fixed points of contact will be. “With a new patient, the parents immediately meet the whole team: dermatologist, nurse, neonatologist and geneticist,” says Dr Colmant.

Those who receive the diagnosis later in life go through a similar pathway too. Patients who have spent years with unexplained complaints finally find clarity at UZ Leuven. “Sometimes we see people who have already been elsewhere, but whose symptoms were not recognised,” says Dr Colmant. “We can often make clear very quickly what it is: this is EB.”

An expertise platform for tailored care

After the diagnosis, the team tailors care completely to the patient. Wendy Godts, EB nurse for adults and care coordinator, and Jill Boeykens and Miet Neyens, EB nurses within the KITES team for children, are the key figures in day-to-day support. Wendy: “I first talk with the patient: what are they up against, what do they need? That helps me decide which specialists we bring into the consultation.”

The EB team

That consultation is a genuine expertise platform. In addition to the dermatologist and the EB nurses, psychologist Joanna Willen, social worker Inge Droesbeke, a dentist, internist, physiotherapist, dietitian, speech therapist and other specialists are involved, depending on the patient’s needs. “EB is far more than a skin disease,” stresses Wendy Godts. “As well as their fragile skin, on which blisters and small wounds quickly appear, some patients also have eye problems, swallowing difficulties, pain, itching… As far as possible, we try to plan everything on one day, so that patients do not have to keep coming back.”

From hospital to home

For younger patients, the KITES team plays a special role: this specialised paediatric liaison team organises care at home. Jill Boeykens: “As well as setting up wound care and pain relief, we also visit schools and liaise with CLBs, community nurses, GPs, physiotherapists and the OCMW… We are available around the clock for all patients and their care providers. Multidisciplinary care therefore does not stop at the hospital door.”

When a new diagnosis is made, families also receive a welcome pack – an initiative of the KITES team, sponsored by DEBRA Belgium vzw – containing dressing material, basic care equipment that is often missing at home but is nevertheless important (a bath thermometer, spatulas, needles to lance blisters, soft bamboo muslin cloths…) and information booklets.

Psychological support

An EB diagnosis comes as a shock to parents and patients alike. Psychologist Joanna Willen supports families through all stages of life: “I like to get to know them during the difficult period of diagnosis, because the situation brings many emotions with it. That creates a bond of trust which really makes a difference in the years that follow.” She supports parents with questions about daily care, helps children with questions about feeling ‘different’, and supports adolescents and adults around themes

For young patients, the family receives a welcome pack that includes, among other things, a child-friendly information booklet.

such as friendships, intimacy and choice of studies. "A chronic disease affects the whole family. Brothers and sisters also deserve extra care and attention."

Social worker Inge Droesbeke also plays an indispensable role. She handles the reimbursement files for the costly dressing material, which can amount to thousands of euros per month, and guides families through the administrative maze. The applications are complex and have to be submitted every year, even though the genetic condition never disappears.

Lifelong support

In some severe forms of EB, palliative support is needed from a young age. The KITES team plays a crucial role in this, including for adult patients whom they have known for years. Jill Boeykens: "We are not strangers who only appear at the end of life. We support people from birth onwards."

"Patients say: 'I feel that you truly know my condition.'"

Patients with severe EB come for follow-up four times a year, while patients with milder complaints come once a year. Ingrid Jageneau from DEBRA Belgium vzw is also always present then, as an expert by experience and patient representative. Dr Colmant: "Patients feel heard and understood here. They often say it themselves: 'I feel that you truly know my condition.'"

The EB team at UZ Leuven is not a theoretical construct, but close collaboration in practice – a team that is there from the first breath through to the final farewell, and everything in between.

Recognition as a centre of expertise

Since 2025, UZ Leuven has been the only hospital in Belgium with an NIHDI convention for EB. Thanks to that recognition, the hospital can organise and fund specialised multidisciplinary care for all Belgian patients with EB. UZ Leuven is also part of ERN-Skin, the European Reference Network for rare skin diseases, and works closely with other European centres of expertise.

GPs, dermatologists and other specialists who see patients with characteristic symptoms can turn to the EB team for diagnosis, advice or referral.

DEBRA Belgium vzw – partner for every EB patient

DEBRA Belgium vzw is the patient organisation for people with EB and their families in Belgium. The association offers practical and emotional support: it organises peer contacts, advocates to the authorities for better support and reimbursement of medicines and care materials, and provides a welcome pack for newly diagnosed patients. In Belgium, the association supports more than 150 families.

The face behind the diagnosis

In 2025, DEBRA Belgium vzw launched an impressive campaign created by photographer and documentary maker Lieve Blancquaert. She portrayed three Belgian EB families, who speak openly about what it is like to live with EB. The campaign, with moving photos and videos, can be viewed on YouTube and Facebook.

More info: debra-belgium.org
View the campaign:
youtube.com/@debrabelgium5574
facebook.com/DebraBelgium

Newborn blood spot screening: a powerful tool for the early detection of rare diseases

Since the 1960s, newborn blood spot screening has been offered systematically and free of charge in Belgium. Much has changed since then. Even in 2026, there is still scope to detect more conditions in newborns and treat them early.

A new Flemish Centre for Neonatal Screening (VCNS)

On 1 January 2026, the organisation and analysis of the newborn blood test were taken over by UZ Leuven and UZ Gent, after previously being carried out in the university hospitals of Brussels and Antwerp. Together, UZ Leuven and UZ Gent set up a new Flemish Centre for Neonatal Screening (VCNS), which coordinates this population screening programme for the whole of Flanders and acts as a central point of contact. The samples are divided between the laboratories of the two hospitals, where they are analysed using the same techniques and standards.

The VCNS also aims to help shape the future of newborn blood spot screening. Using the standard methods of biochemical or targeted genetic tests, it is possible to detect still more diseases, something in which some other countries are already further ahead. Thanks to new technologies, a further possible step is now emerging: genomic neonatal screening. By analysing a newborn's full DNA, conditions can be detected that remain invisible with current tests.

What is newborn blood spot screening?

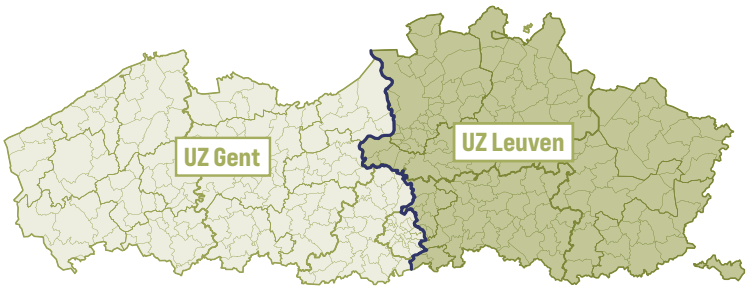
- ✓ Also known as the population screening programme for newborns, neonatal screening, the Guthrie test or the heel-prick test.
- ✓ Between 48 and 96 hours after birth, with the parents' consent, a small blood sample is taken from the baby by means of a prick in the hand and placed on a card.
- ✓ Biochemical or targeted genetic tests can then detect various congenital rare diseases.
- ✓ At present, the following diseases are screened for in Flanders:
  - 14 inherited metabolic diseases
  - cystic fibrosis
  - 2 hormonal disorders
  - spinal muscular atrophy
  - from mid-2026 onwards, severe combined immunodeficiency will also be detected through a targeted genetic analysis.

These are all diseases that can be treated. The earlier they are detected, the sooner treatment can begin (for example medication or a diet). For several of these diseases, speed is essential. In metabolic diseases, for example, certain nutrients are toxic for the child. An early diet can prevent a build-up of harmful substances in the body.

Towards genomic neonatal screening

Extending newborn blood spot screening with genomic analysis offers many possibilities, while at the same time raising important questions. Genetic information is not black and white: not every genetic variant leads to disease, and the course can vary greatly from one child to another. That is why the Belgian university hospitals and genetic centres jointly developed a national vision:

- ✓ **Embedding in care.** Genomic neonatal screening must be embedded in a broader vision of genetic care across the whole life course. Clear information for current and prospective parents, and careful support, are indispensable. Parents need to know what screening can and cannot mean, and that genetic screening does not always provide a definitive diagnosis or rule out all congenital diseases.
- ✓ **Data management and privacy.** Genomic data are very large and must be properly protected. A secure infrastructure for data storage and clear guidelines on who may consult which data and when are essential.
- ✓ **Equal access.** Screening must be accessible to all parents within public healthcare, regardless of place of residence, background or financial means.
- ✓ **Complementary to the current approach.** Genomic screening does not replace the current newborn blood test but complements it. Even within the current test, a number of diseases that are easy to detect and treat could already be added urgently. Biochemical analysis can also help interpret genomic data and is, moreover, ultra-fast, which is necessary for some diseases in order to avoid harm.



The blood spot samples are divided between the laboratories of UZ Gent and UZ Leuven.

The path towards genomic neonatal screening must therefore proceed step by step, through a carefully designed national project that pays attention to medical, ethical, organisational and societal aspects. In this way, we are building a future in which more children with a rare disease receive the right care in time, within a strong and solidarity-based health system.

On 12 February 2026, the national vision text on genomic neonatal screening was presented at a policy forum organised by the pharmaceutical consortium RADDIAL and the patient association RaDiOrg, together with the Belgian university hospitals.

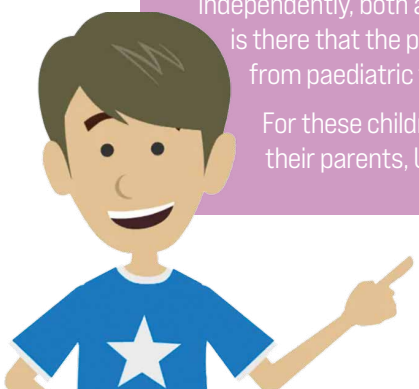
Read the full vision text on genomic neonatal screening here: qr.uzleuven.be/gNBS

E-learning on transition

When a child gradually grows into adulthood, that is in itself an exciting and eventful period. For children and young people with a chronic disease, transition is added to this. In a nutshell, transition means managing the disease progressively more independently, both at home and in hospital. It is there that the patient ultimately moves from paediatric to adult care.

For these children and young people and their parents, UZ Leuven has developed four interactive e-learning modules that provide extra support during the transition process in the form of tips and tricks, personal testimonies and information on all kinds of themes. Each e-learning module is aimed at a specific target group (12-13 years, 14-15 years, 16-18 years and parents).

The e-learning is available free of charge in Dutch via:



Data as the key to better care and policy

Reliable data are essential for making well-founded decisions about healthcare policy. That is why the Central Registry of Rare Diseases, managed by Sciensano, collects crucial data on the diagnosis, demography and care pathway of patients with a rare disease in Belgium. This register not only makes it possible to better understand the epidemiology, but also supports care, research, policy and access to therapeutic innovation.

The register was set up in 2012 and initially collected genetically confirmed diagnoses from the Belgian genetic centres. Since 2025, the rare diseases functions have also been reporting to the register. In this way, the register is gradually growing into as complete an overview as possible of all rare disease diagnoses in our country. The new Belgian Rare Diseases Plan also provides additional guidelines and resources to further expand it and connect it to other national and European initiatives around data sharing, policymaking and care organisation.

Automated registration at UZ Leuven

To participate in this register, UZ Leuven developed a system that automatically collects and transmits data from the electronic patient record. The data are pseudonymised, so that the patient's identity remains concealed.

This system forms part of a broader move towards more structured registration in the patient record: each disease is assigned a unique code linked to the patient, and this code can be easily extracted for reporting and research without extra administrative burden for the healthcare professional. At present, we are training our doctors to use this registration as a routine part of care. The aim is that, in time, all diagnoses will be recorded and reported in this standardised way.

More information on the Central Registry of Rare Diseases: www.sciensano.be/en/projects/central-registry-rare-diseases

Rare diseases in KU Leuven's Master's in Medicine

The rapid recognition and referral of patients with a rare disease, or suspected rare disease, starts with the alertness of every doctor. From the 2026-2027 academic year onwards, rare diseases will therefore have a fixed place in KU Leuven's Master's in Medicine.

- ✓ In **phase 1** of the master's programme, from 2026-2027 onwards students will be able to choose a multidisciplinary elective introducing them to the specific challenges and organisation surrounding rare diseases. Using case studies, they will learn how to recognise suspected rare disease more quickly, how to refer efficiently, and what role centres of expertise play in further diagnosis and follow-up.
- ✓ In **phase 3** of the master's programme, from 2028-2029 onwards the elective *Integrative organisation and multidisciplinary treatment of patients with rare diseases* will be offered. It explores diagnosis, treatment, care organisation and the policy framework in more depth (e.g. orphan medicines).

The course units were developed by experts from UZ Leuven. In this way, we want to help shape a new generation of doctors who are better prepared to recognise and support patients with a rare disease.

Leuven.IRD is building an ecosystem that connects research, care and societal needs in rare diseases

Rare diseases are individually rare, but far from insignificant for society. For many patients, the reality remains stark: a diagnostic odyssey averaging five years, frequent misdiagnoses, and for 95% of conditions there is still no effective treatment. To respond to that challenge, Leuven.IRD brings together the scientific expertise of KU Leuven and UZ Leuven in an integrated ecosystem. Its aim is to stimulate knowledge, awareness, innovative research and therapy development for rare diseases, ultimately helping people with a rare disease.

Leuven.IRD as a catalyst for rare disease research

Generating new insights into rare diseases is not only a scientific challenge, but also faces structural barriers. Bottlenecks include structural underfunding, expertise that is hard to locate, the slow translation of fundamental research into clinical applications, underuse of valuable patient data and limited insight into patient needs.

Leuven.IRD explicitly starts from these bottlenecks and develops solutions to address them structurally. To achieve this, the institute focuses on **four pillars** in scientific research.

1. **Making expertise visible and accessible**
   KU Leuven and UZ Leuven have broad expertise in rare diseases, spread across research groups, faculties and clinical departments. Leuven.IRD brings this expertise together through

Leuven.IRD was officially inaugurated on 24 September 2025.

a dynamic expertise database and a central contact point, so that researchers, clinicians and external partners can find the right expertise more quickly and new collaborations can arise more easily.

2. **Integrating clinical and genetic data**
   Although UZ Leuven has clinical and genetic information on tens of thousands of patients with rare diseases, its potential remains underused. Together with the hospital, Leuven.IRD wants to invest in an integrated and future-oriented data infrastructure. This includes standardising clinical data and making data available for research within a robust ethical and legal framework that guarantees proper use and patient privacy. In this way, we create opportunities to generate new insights that can lead to faster diagnosis and new treatment options.

3. **Putting the patient and their needs at the centre**
   Patient needs go beyond medical treatment alone and also include psychosocial support and access to information and care coordination. Leuven.IRD will identify, measure and translate the needs of patients with rare diseases into concrete recommendations for researchers, healthcare professionals and policymakers. The aim is to align care and innovation more closely with what patients really need.

4. **Accelerating diagnosis and therapy**
   Small patient groups, complex regulation and fragmented collaboration often slow research and its translation into clinical applications. By stimulating multidisciplinary collaboration, lowering barriers to innovation, setting up targeted flagship projects and investing in awareness and training, Leuven.IRD wants to act as an accelerator for diagnosis and therapy development and speed up the translation of fundamental research into clinical impact.

Raising public awareness of rare diseases

Despite the large number of patients, rare diseases often remain under the radar. The lack of knowledge still too often leads to misunderstanding and underestimation. Through public activities, educational initiatives, lectures and workshops, often in collaboration with other organisations, Leuven.IRD brings rare diseases closer to society. For example, on the Dag van de Wetenschap Leuven.IRD offers tours of research laboratories, organises workshops on rare diseases for young and old, and raised money for De Warmste Week through Race for Rare.

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Zicht op Zeldzaam: sharing knowledge and building connections

On Rare Disease Day 2026, Leuven.IRD launched the **Zicht op Zeldzaam** (Insight into Rare) platform. There you will find reliable and accessible information: from easy-to-understand blogs, patient testimonies and educational videos to the podcast series Under the Radar. In that podcast, patients and scientific experts talk together about relevant themes. The aim is not only to inform, but also to connect: bringing patients, healthcare professionals and researchers closer together and increasing societal understanding of rare diseases.

Discover the platform: kuleuven.be/ird/insight-into-rare

laboratories, organises workshops on rare diseases for young and old, and raised money for De Warmste Week through Race for Rare.

Rare diseases confront us with the limits of traditional structures in care and research, while at the same time acting as a powerful engine for innovation, with insights that are often applicable far beyond rare disease alone. Leuven.IRD consciously opts for a model in which collaboration is central: across disciplines, institutions and sectors, with active involvement of patients. In this way, Leuven.IRD is building a future in which rare no longer means invisible, misunderstood or untreated.

Call for input

Leuven.IRD is an open and growing initiative. Do you have any ideas, suggestions or themes that we could include in our work or podcasts? Would you like to share a testimony or contribute actively to the institute?

You can contact us via

- ✓ rarediseases@kuleuven.be
- ✓ kuleuven.be/ird
- ✓ linkedin.com/company/leuven-ird

Honorary doctorate for rare diseases

During KU Leuven's Patron Saint's Day celebration, Professor Ségolène Aymé (°1946) received an honorary doctorate this year on the nomination of Leuven.IRD. Her visionary work as a medical geneticist and epidemiologist has had a profound impact on scientific research and care for rare diseases, in France and across Europe. For Leuven.IRD, Ségolène Aymé's work is a source of inspiration.

Aymé is emeritus research director at the French National Institute of Health and Medical Research (INSERM) and is attached to the Hôpital de la Salpêtrière in Paris as a rare diseases expert. In 1997 she founded Orphanet, which under her leadership grew into Europe's largest knowledge platform for rare diseases. As a member of numerous European and international initiatives, including the WHO advisory committee on rare diseases, Aymé

has campaigned to bring rare diseases to wider attention in Europe and worldwide. In 2004 she was the driving force behind the first French plan for rare diseases. This now forms the basis of a pan-European care system, the European Reference Networks for rare diseases (ERNs), in which centres of expertise are brought together so that patients can receive the right specialised care.

Support the Fund

By contributing to the Rare Diseases Fund, you can support scientific research and care for people with a rare disease.

Professor Gert Van Assche, Medical Director of UZ Leuven and manager of the fund: "For patients with rare diseases, research into disease mechanisms and new treatment options is extremely important, as there is often still no adequate therapy. KU Leuven and UZ Leuven wish to pool their unique expertise to offer precisely these patients and their children a better future. Your support for this research can truly make a difference."

More information about the fund and donations can be found on the Rare Diseases Fund website: qr.uzleuven.be/bgGB9R

OVERVIEW OF USEFUL LINKS

UZ Leuven rare diseases

www.uzleuven.be/en/rare-diseases

- ✓ In which situations and through which routes can patients turn to UZ Leuven in case of suspected or diagnosed rare disease?
- ✓ More information about the teams specialised in diagnosing and treating rare diseases
- ✓ News and events

Registration form in case of (suspected) rare disease

Patient registration form via www.uzleuven.be/en/rare-diseases or directly at qr.uzleuven.be/bgGB0j

Referring doctor registration form via www.uzleuven.be/en/rare-diseases or directly at qr.uzleuven.be/bgGAs9

Support the Rare Diseases Fund with a donation: qr.uzleuven.be/bgGB9R

E-learning transition from paediatric to adult care (in Dutch): www.uzleuven.be/kindergeneeskunde/onderweg

Leuven Institute for Rare Diseases (kuleuven.be/ird)

- ✓ Brings together expertise in rare disease research within KU Leuven and UZ Leuven

Flanders

Vlaams Netwerk Zeldzame Ziekten (Flemish Rare Diseases Network – www.departementzorg.be/vlaams-netwerk-zeldzame-ziekten)

- ✓ Organisation in which Flemish university and general hospitals, GP circles, and numerous patient associations join forces.

Belgium

NIHDI (www.riziv.fgov.be)

- ✓ NIHDI agreements: via *Thema's* > *Verzorging: kosten en terugbetaling* > *Ziekten*, you can see for each disease which care the health insurance fund reimburses.

RaDiOrg (www.radiorg.be)

- ✓ The Belgian umbrella association for people with a rare disease. RaDiOrg brings together more than 80 associations for specific rare diseases as well as hundreds of individual members with a disease for which no association exists. RaDiOrg is the national alliance of EURORDIS, the European federation for rare diseases.

Europe

Orpha.net (www.orpha.net)

- ✓ International information portal on rare diseases and orphan medicines, aimed at patients and professionals. Orphanet manages the nomenclature and classification of rare diseases, and publishes for each disease the reference centres recognised in Europe for diagnosis and treatment. For Belgium, you can identify the centres of expertise for each specific disease by this logo (without the additional RC symbol):

European Reference Networks (health.ec.europa.eu/rare-diseases-and-european-reference-networks_en)

- ✓ Overview (from the European Commission) of the European Reference Networks for rare diseases, within which members exchange knowledge and information.

European University Hospital Alliance (EUHA) (www.euhalliance.eu)

- ✓ Network of eleven major university hospitals in Europe. A specific working group is handling a number of improvement projects on rare diseases.

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