Lucie Albon

From A to Rare diseases

in collaboration with
Prof. Laurence Faivre, Élodie Gautier
and Sonia Goerger

FONDATION IPSEN
BookLab

Lucie Albon

Rare Diseases
from A to Z

in collaboration with the
Platform of Expertise for Rare Diseases – Bourgogne-Franche-Comté

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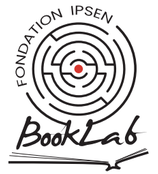


This pictogram, which is present on certain pages, redirects to transcriptions, done by Sonia Goerger, of testimonials from patients suffering from the disease presented or from their entourage (in French) – **www.pemr-bfc.fr**.

Thanks to the collaboration of Ewen Life **www.ewenlife.org**, this pictogram, displayed on certain pages, redirects to video testimonials of patients affected by the disease presented (in French).

How to tackle the illustration of a disease?

By Lucie Albon

After creating wall art in the rooms of the intensive care unit at the Chalon-sur-Saône hospital and a painting project for the Woman-Mother-Child hospital (HFME) in Lyon, the dialogue between art and disease was initiated. I started this new challenge with the experience of my first creations in mind.

When I visited the intensive care unit, the rapport I had with the patients was immediate. Many conversations took place and the feedback I received helped me to progress. I understood the importance of creating images that were "open to interpretation", where everyone can access the artwork through the door that suits them best.

What I have tried to do here is to be both meaningful and abstract. It is possible to see meaning in the following images - at least I hope so, because that was my intention - but the reader, the viewer, is not obliged to do so. One can also simply be transported by the image.

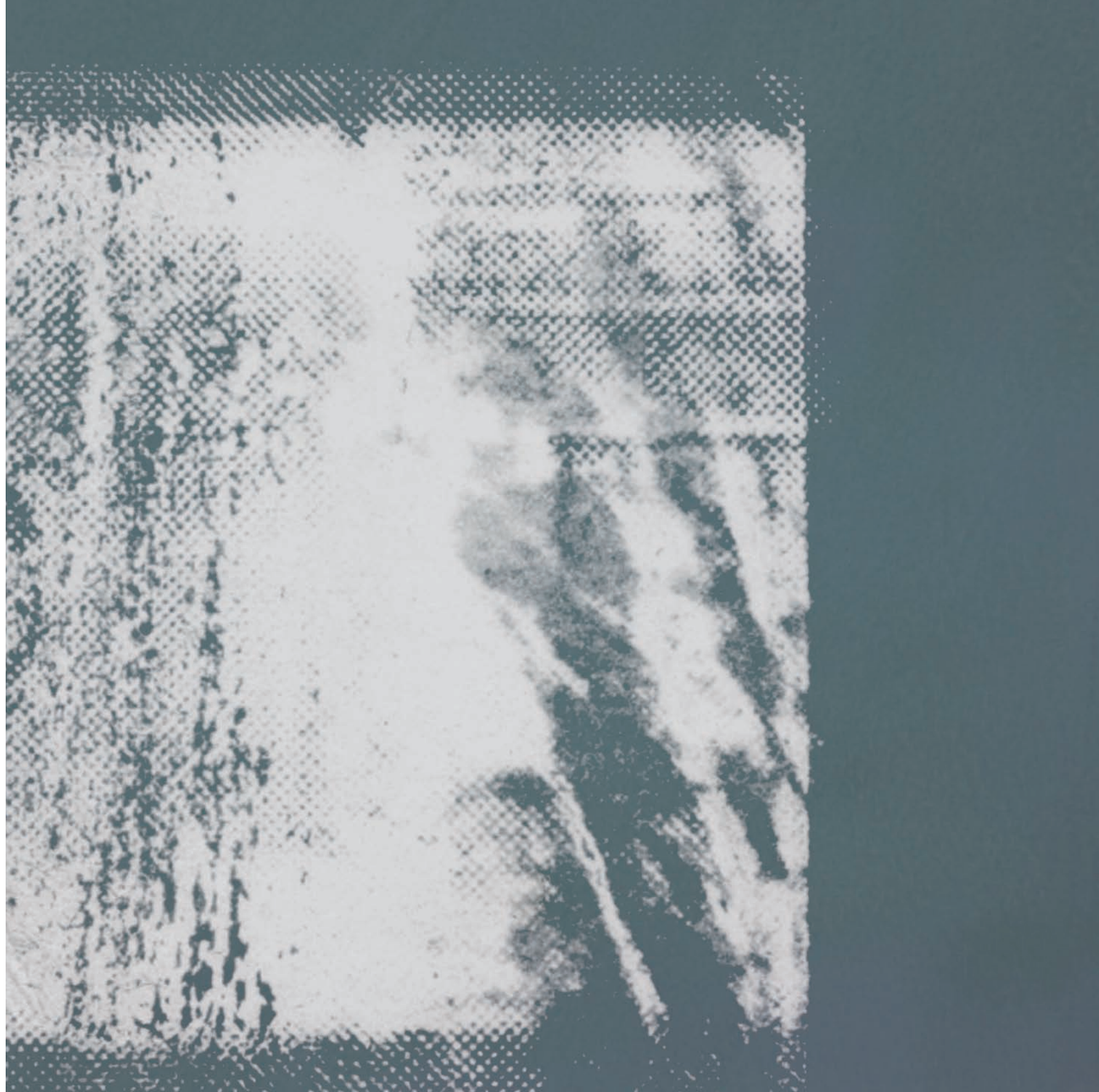
The discovery of an illness in a loved one, be it rare or not, is difficult to accept. The idea of this alphabet book is to demonstrate that there are a multitude of rare diseases and that we are not alone in facing this challenge.

I used my emotions to illustrate, instinctively, each of these diseases. I tried to remain coherent while at the same time allowing myself a lot of freedom. This work is a mixture of different visual art techniques: painting, ink drawing, decoupage, digital tools, stencils, linocuts, monotypes, prints, graphic design. In designing these works of art, I imagined that they would reflect the disease description. The "text-image" relationship was further enhanced through the collaboration with graphic designer Matthieu Perret. Today, I believe that this collaborative partnership is essential.

I hope that this creation will be an entry point for families, caregivers and associations and will provide a medium to allow for dialogue and to raise public awareness.

AMYOTROPHIC LATERAL SCLEROSIS

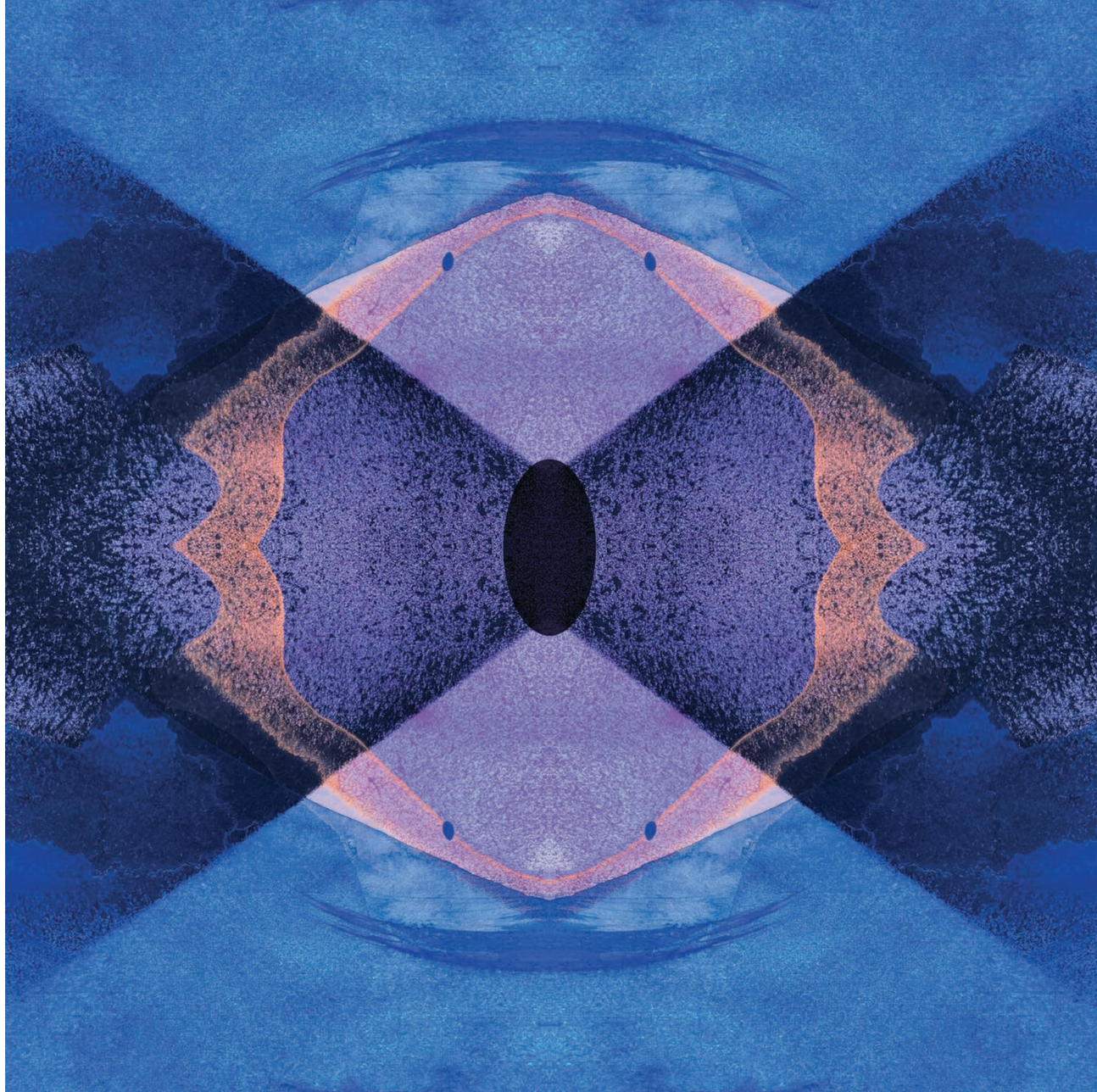
Amyotrophic lateral sclerosis (ALS) or **"Lou Gehrig disease"** is a **progressive and fatal neuromuscular disease** characterized by a loss of motor neurons, the neurons which control, among other functions, our walking, speaking, swallowing and breathing. It is characterized by a **progressive death of motor neurons**, muscular atrophy and therefore the progressive paralysis of patients. The mean age of onset is approximately 60 years, but may be earlier, especially in familial cases (5-10% of cases). The disease is most often fatal within two to five years after onset. To date, there is one drug that has been shown to **slow the progression** of the disease and an **experimental therapy** using an antisense oligonucleotide (a fragment of RNA, usually synthesized in the laboratory that can specifically bind to the natural messenger RNA that it targets) is providing hope in the research setting.



b

BEHÇET

Behçet disease is an **inflammatory disease of the blood vessels**. It is multifactorial disease, with both genetic and environmental risk factors. It manifests mainly as **ulcers in the mouth** or on the genitals, but also with eye, skin, joint, and nervous system involvement and venous or arterial occlusions. Severe fatigue is also reported. It mainly affects young people, with age of onset most often being between the ages of 10 and 45 years. It is a chronic disease which evolves with attacks of varying intensity. Ultimately, the prognosis is primarily related to the risk of sequelae (particularly ocular and neurological) in the case of severe attacks. Several **treatments** are available to reduce the signs and symptoms of the disease, including corticosteroids.

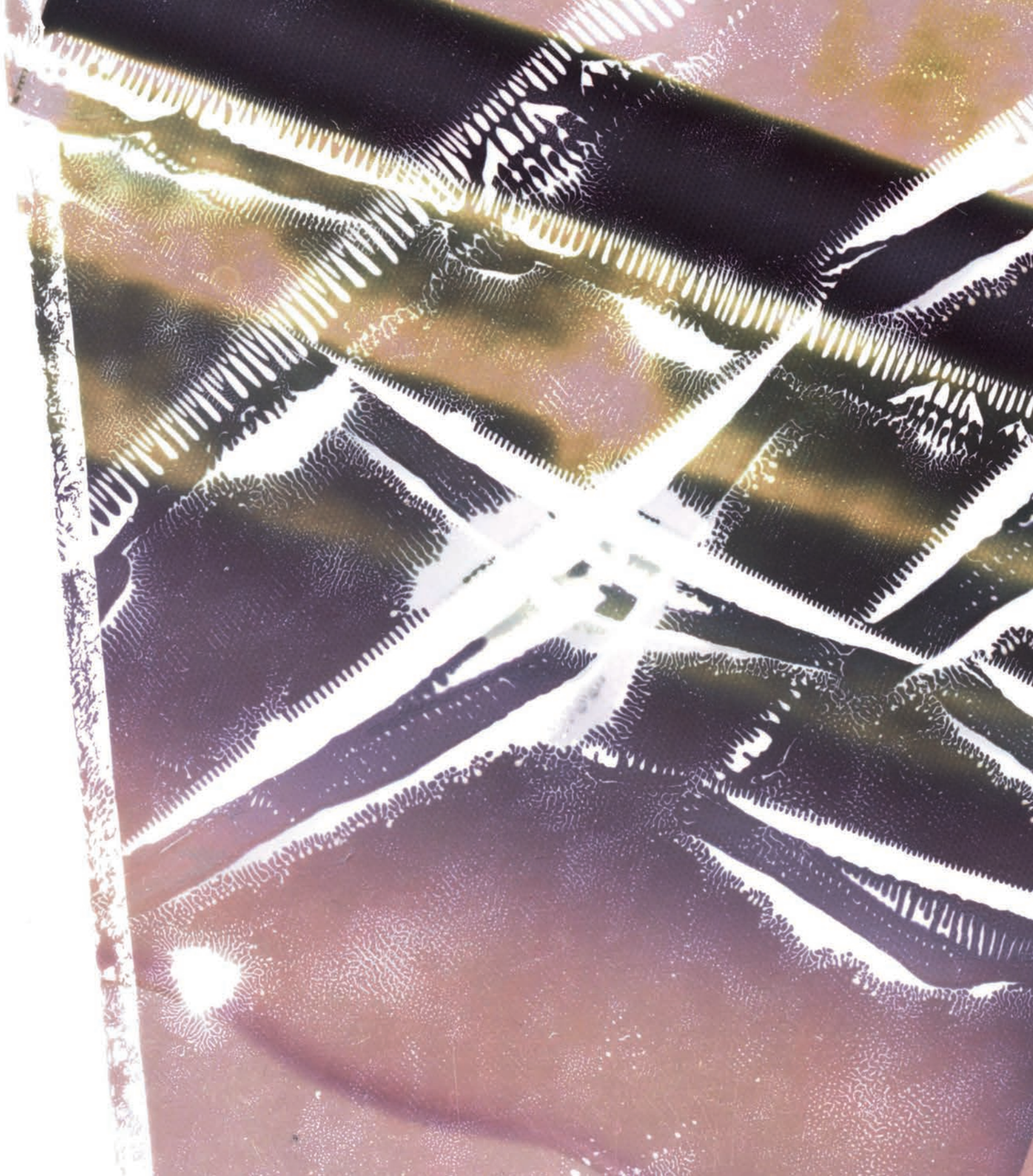


CYSTIC FIBROSIS

Cystic fibrosis is the most common inherited genetic disease. It is passed on to offspring when **both parents** carry a variant in the *CFTR* gene. It mainly affects the **lungs**, but also the d**igestive and reproductive systems**. It is the degree of respiratory damage, characterized by bronchial dilation, which reveals the seriousness of the disease and determines the prognosis. This disease is screened for at birth. Before the arrival of revolutionary treatments, it was fatal in young adulthood and required regular follow-up and management involving respiratory physiotherapy. Today, **treatments** allow affected patients to lead an almost normal life, with a significant reduction in the management required. The treatments are, however, not effective in a small number of patients with specific mutations.

DUCHENNE MUSCULAR DYSTROPHY

Duchenne muscular dystrophy (DMD) is a genetic disease that mainly affects boys as the causal gene is located on the **X chromosome**. The disease presents in early childhood, with problems **walking**, difficulty **climbing** stairs and frequent **falls**. Patients lose their ability to walk between the ages of 6 and 13. Those who do not receive corticosteroids die between late adolescence and young adulthood from **respiratory failure** and/or **cardiac hypertrophy**. Certain treatments can be given based on the particular mutations found in the gene, which is referred to as **personalized medicine**.



EVANS

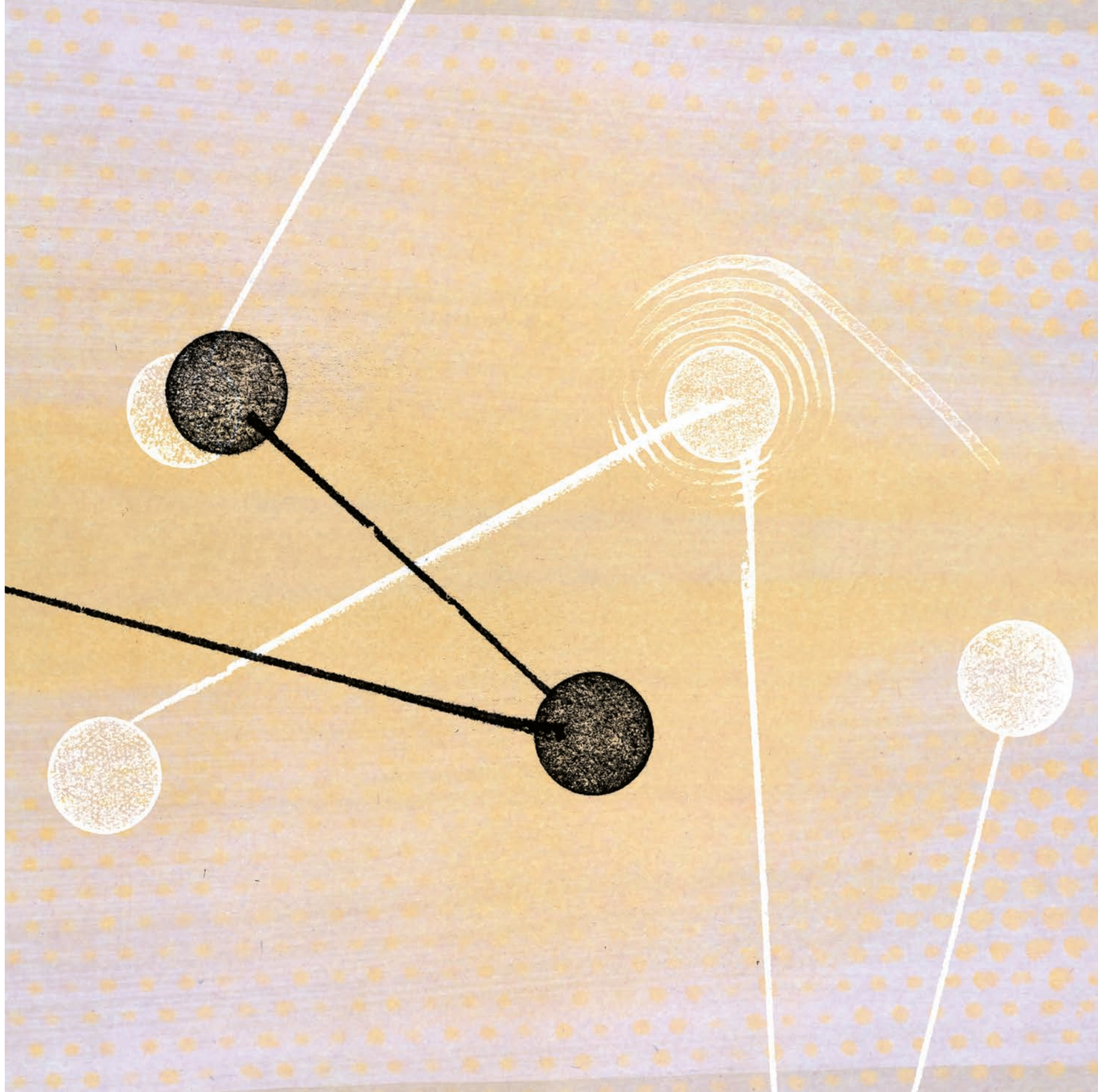
Evans syndrome is a rare hematological disorder, which means a disease that affects the blood. This condition is also described as an **autoimmune** disease because the body's immune system turns against certain of its own vital elements. In the case of Evans syndrome, agents of the immune system attack several components of the blood including red blood cells and platelets. The syndrome can occur in childhood or in adulthood. During attacks, the consequences are **anemia** leading to fatigue and/or a decline in platelet counts that can result in episodes of bleeding. It is a chronic disease with alternating periods of remission and relapse that can sometimes, despite **treatment**, be fatal due to severe hemorrhage or infections. Corticosteroids or immunosuppressive treatments are necessary during an attack.

PULMONARY FIBROSIS

Idiopathic pulmonary fibrosis (IPF) is a **chronic** lung disease characterized by the progressive alteration of the lung **structure** in the form of irreversible scarring. IPF generally presents after the age of 50 and mainly in men. The cause of the disease is not yet known. Manifestations include **progressive shortness of breath** and a **dry cough**, which can progress to premature death in some patients in the absence of lung transplantation. Today, there are medications that can improve symptoms. It is crucial for smokers to stop the use of tobacco.

GITELMAN

Gitelman syndrome is a genetic disease that causes an abnormality in the kidneys, leading to the excessive elimination of potassium. It is passed on to offspring when **both parents** are carriers of a variant in the gene responsible for the syndrome. It manifests mainly in teenagers and adults, but also sometimes during childhood. It causes **hypokalemia** (a decrease in the concentration of potassium in the blood) and a tendency for the blood to become more alkaline (**alkalosis**), which in turn leads to **hypocalcemia** (low calcium levels in the blood).
The associated signs are primarily **muscle pain and cramps**, **dizziness**, **delayed growth**, episodes of **tetany** and **fatigue**. Symptoms may improve when patients are given salt, potassium and magnesium supplementation.

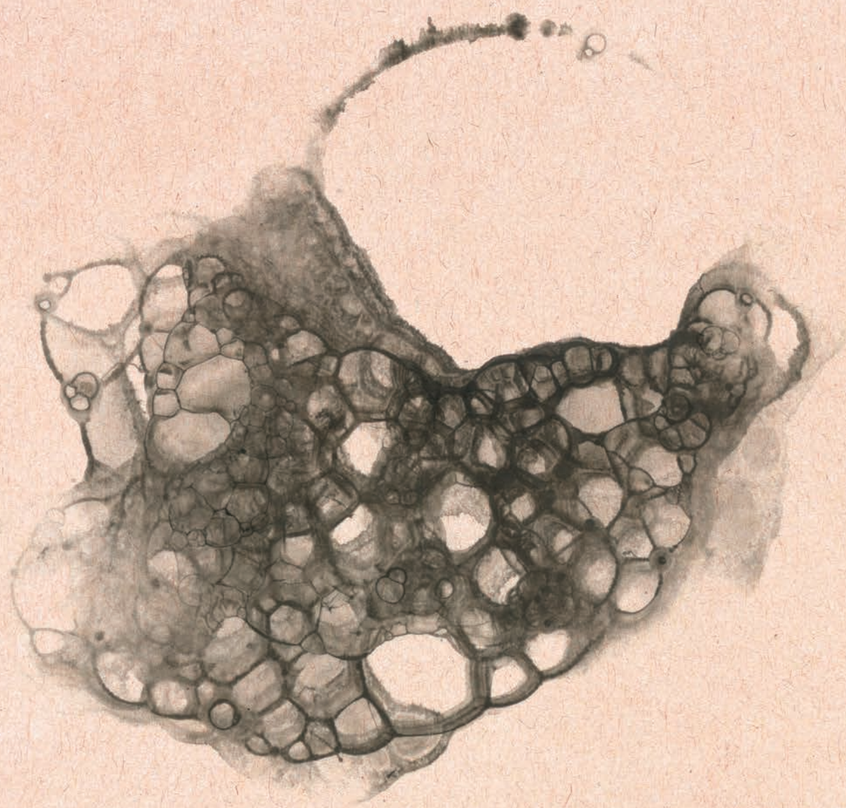


HEMOPHILIA

Hemophilia is a **hereditary** disease caused by a deficiency of **coagulation** factors VIII or IX. The causative gene is located on the **X chromosome**. Therefore, women who have little or no signs because they have two X chromosomes can pass the disease on to their sons who have only one X chromosome. It is diagnosed very early in boys with a severe deficiency and later in childhood in boys with a mild deficiency, or in girls. In a family where the disease is known, it is most often diagnosed during pregnancy, or at birth. It results in the failure of blood to coagulate: when bleeding occurs, it cannot be stopped without treatment or without great difficulty. The consequences are significant, with **severe hemorrhaging** following an injury and **spontaneous bleeding** sometimes occurring, particularly in the joints or the brain. If left untreated, joint deformation and physical disability can develop. **Recombinant factors**, which are **biologically engineered** factor VIII products, have been around for a long time, and can be administered following a bleeding episode or to prevent bleeding. Other treatments, including **gene therapy**, are currently being developed.

INCONTINENTIA PIGMENTI

Incontinentia pigmenti is a genetic disease that occurs essentially in females and which mainly affects the **skin**, **teeth**, **eyes** and central **nervous system**. It is caused by a mutation of the *NEMO* gene, located on the **X chromosome**. When passed on to a male fetus, with only one X chromosome, it results in a spontaneous abortion of the fetus. Diagnosis is most often made based on the presence of characteristic skin signs (appearance of blisters at birth, later replaced by plaques and then pigmented patches that persist throughout life). When it is limited to the skin, the disease is most often benign, but some women can have serious **neurological and ophthalmological complications**. There is no specific treatment, but **management for the various symptoms** exists.



j

JEUNE

Jeune syndrome, also known as asphyxiating thoracic dystrophy, is a rare disorder characterized by short ribs, **narrow thorax**, **short arms** and legs and **skeletal abnormalities**. Short stature of varying degrees may be observed. **Intellectual development is normal**. It is transmitted when **both parents** are carriers of a variant in one of the genes responsible for the syndrome. The diagnosis is often suspected during pregnancy, when skeletal abnormalities are detected. Some patients may develop severe respiratory failure, while others have a benign clinical course. There is no specific treatment, but **management is available to address the different symptoms**.

KLINEFELTER

Klinefelter syndrome is a sex chromosome abnormality in which **boys are born with two X chromosomes and one Y chromosome** (XXY) instead of one X chromosome. Affected boys are often **tall** with **long arms and legs**, and may exhibit **learning disabilities**. They present with small testicles, incomplete puberty in the absence of treatment, and are **sterile**. The diagnosis is most often suspected at puberty and **treatment with testosterone** may be beneficial. Carrier couples can be referred to assisted reproduction centers if they wish to have children. This condition is above the frequency threshold for a rare disease in Europe, but certain **variants with more than 3 sex chromosomes**, which have different consequences, are very rare.

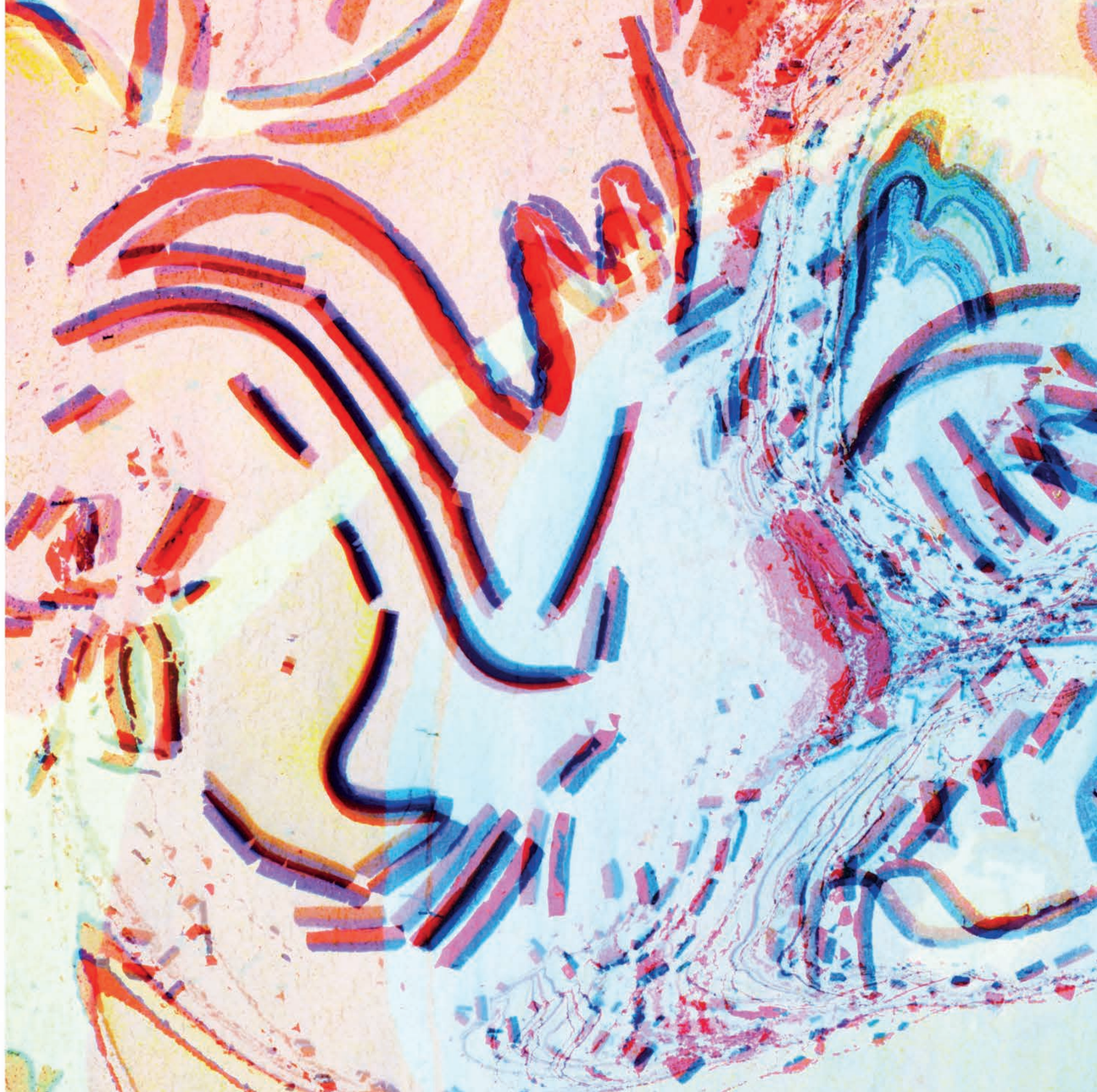
1

LOW PHOSPHOLIPID-ASSOCIATED CHOLELITHIASIS

Low phospholipid-associated cholelithiasis (LPAC) is a **genetic predisposition** to the formation of cholesterol **gallstones** in, and outside of, the liver. It manifests as intense and intermittent **abdominal pain**, related to the presence and/or migration of gallstones. It often develops before the age of 40, sometimes at the end of or after pregnancy in women. The mode of transmission is referred to as autosomal dominant, meaning that an affected person has a one in two chance of passing on the causative mutation to his or her offspring. **Preventive drug therapy** can be provided in order to avoid the recurrence of symptoms; it is sometimes necessary to remove the gallbladder when the gallstones are causing complications. If left untreated, some patients may develop severe **liver disease** that may require liver transplantation.

MARFAN

Marfan syndrome is characterized by a progressive **dilatation of the aorta** associated with an increased risk of **aortic rupture** (which can be lethal in the absence of emergency surgery); dislocation of the ocular lens; **skeletal signs** with tall stature, long fingers, arms and legs, scoliosis, and chest deformity; stretch marks; and risk of pneumothorax. The age of onset of these signs can vary. Marfan syndrome is caused by a mutation in the *FBN1* gene, and a person with Marfan syndrome, who generally has an affected parent, has a 50% chance of passing the disease on to their children. With regular follow-up and proper management, patients have a life expectancy close to that of the general population, as **life expectancy has increased by 30 years over the past 30 years**.



NARCOLEPSY

Narcolepsy-cataplexy, or **Gélineau disease**, is a rare sleep disorder characterized by **sudden bouts of sleepiness** during the day, possibly accompanied by **cataplexy**, which is a sudden loss of muscle tone. Onset occurs in childhood or young adulthood. It can hinder academic and professional performance, and driving may be contraindicated. Taking **psychostimulants** may improve symptoms. An **autoimmune origin**, resulting in the decrease of a neurotransmitter in the brain, is suspected.

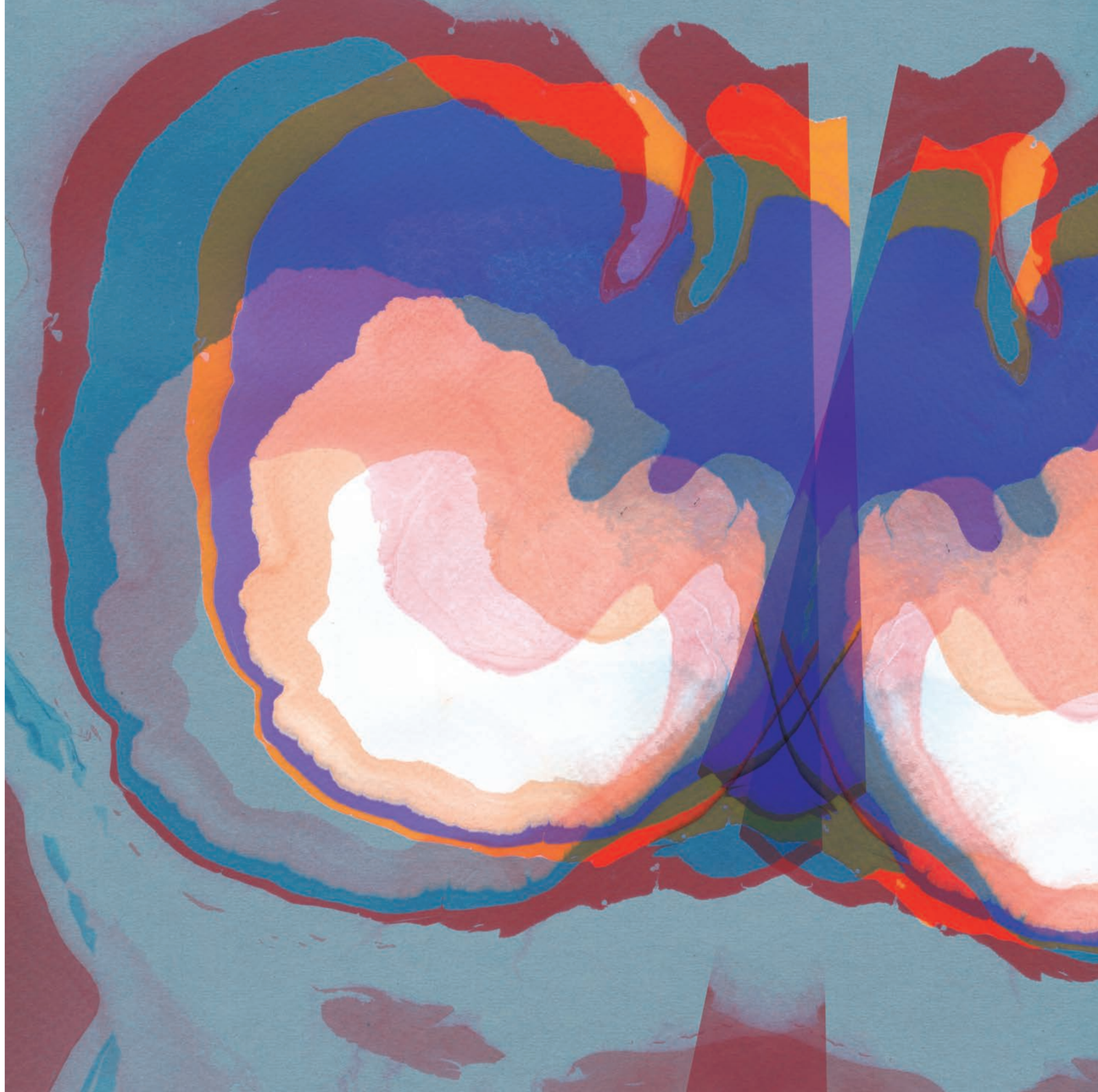
OEIS

OEIS complex is a rare syndrome, of unknown etiology, that associates an **omphalocele** (incomplete closure of the abdominal wall), **bladder exstrophy** (bladder exposed outside of skin), an **imperforate anus** (anus that is not open, preventing the excretion of feces and requiring urgent surgery), and **spina bifida** (a defect in the closure of the lower spine). It is diagnosed during pregnancy, and can often lead to the termination of pregnancy due to the **multiple malformations** and their poor prognosis.

p

PIERRE ROBIN

Pierre Robin sequence is a rare syndrome that is diagnosed during pregnancy or at birth. It associates **retrognathia** (posteriorly placed chin) and **glossoptosis** (posterior displacement of the tongue), with or without **cleft palate**, which can lead to difficulties in swallowing and obstructive breathing problems in neonates. When isolated, this sequence has a good prognosis and is a simple developmental anomaly. Nevertheless, in 50% of the cases, it is **associated with other anomalies** and their association is at the origin of more complex genetic syndromes. Management includes a combination of surgery, particularly to repair the cleft, as well as management of all symptoms, together with nutritional support.



q

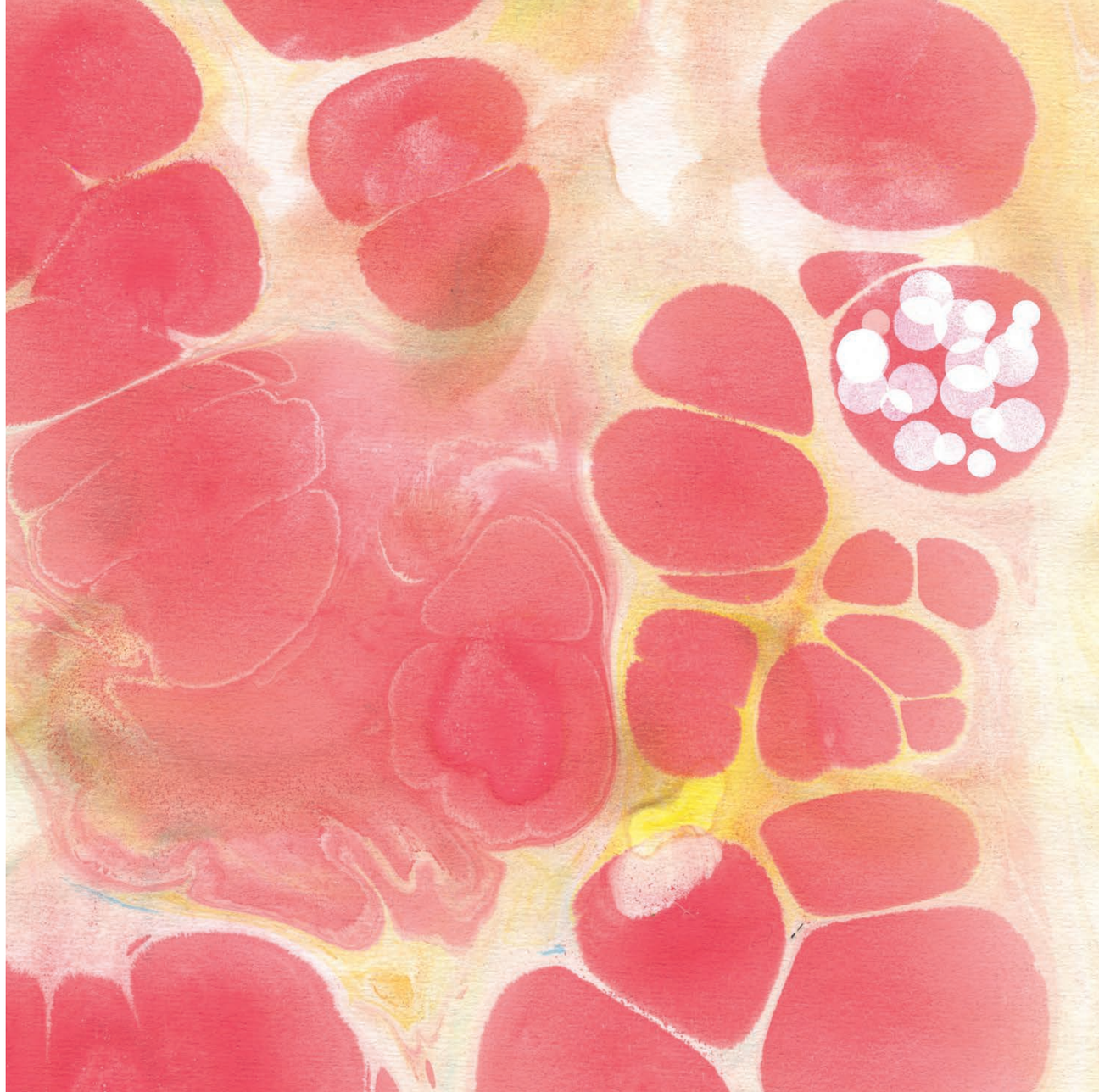
LONG QT

Long QT syndrome is an abnormality of the heart's electrical system, which controls both **heart rate (speed) and rhythm**. When functioning properly, the heart beats at a normal rate and rhythm. When this is not the case, we have what is called an arrhythmia. Long QT syndrome is a type of arrhythmia. It can be caused by certain medications, but it can also be **inherited** from parent to child. The diagnosis is often made in adulthood, although it can sometimes be diagnosed in childhood or even in the neonatal period.
Treatment with medication or a **defibrillator** is required to control arrhythmias, which can lead to fainting or sudden death at any age.

R

RENDU-OSLER

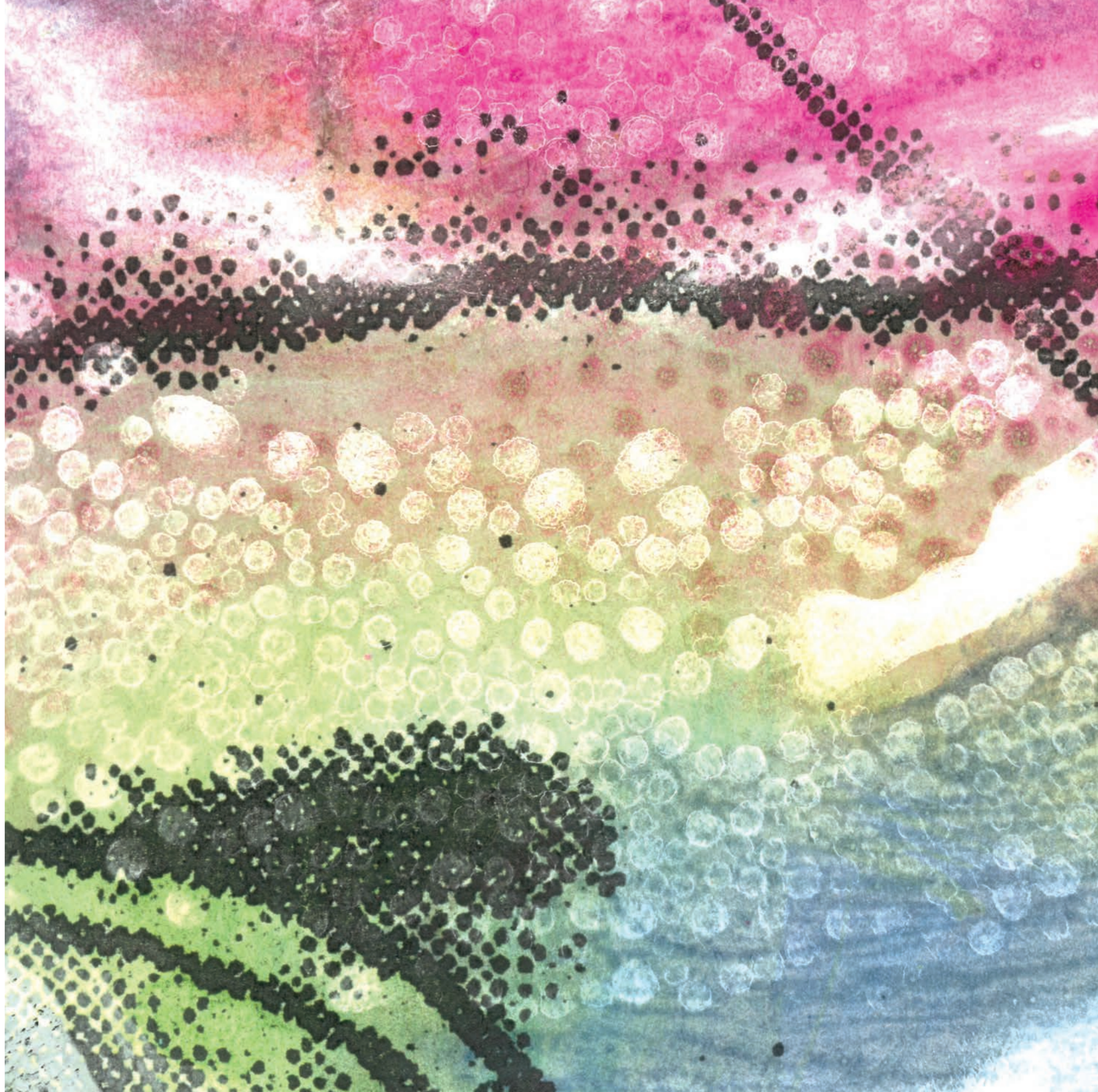
Rendu-Osler disease, more commonly known as **hereditary hemorrhagic telangiectasia (HHT)**, is a disease of the blood vessels characterized by **hemorrhages** (especially nosebleeds), small dilated blood vessels on the surface of the skin and mucous membranes, and **arteriovenous malformations** (which can be located in the lungs, liver and other organs). It is an **inherited** disease that can affect both men and women, with an affected person having a 50% risk of passing on the disease to their children. Diagnosis is based on the presence of **epistaxis** (nosebleeds), the detection of characteristic **skin lesions**, **visceral involvement**, and the hereditary nature of the disease. It can be confirmed by genetic testing by searching for variants in the various genes responsible for the disease. Although the life expectancy of unscreened patients is reduced, the life expectancy of patients who have their arteriovenous malformations managed by a specialized expert center is comparable to the general population. **Treatment** consists of prevention and management of epistaxis and associated anemia, as well as the screening and management, usually with interventional radiology, of arteriovenous malformations.

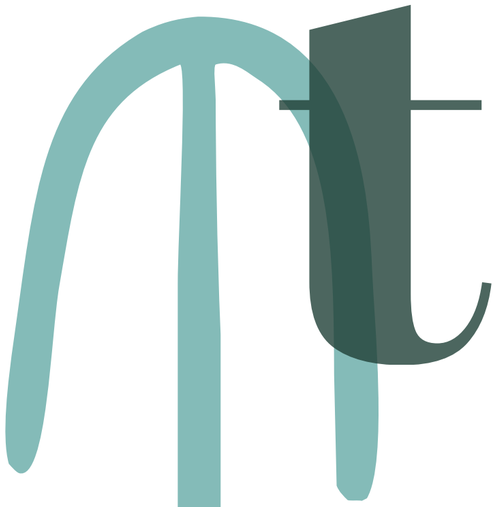


S

SICKLE-CELL ANEMIA

Sickle cell anemia is a genetic disease affecting **hemoglobin**, a substance found in red blood cells responsible for carrying oxygen throughout the body. It leads to the **deformation** of red blood cells. It is passed on to offspring when both parents carry the same alteration in the HbS gene, and is much more prevalent in Africa. Manifestations of the disease include **anemia** (i.e. low levels of red blood cells, resulting in fatigue, dizziness, shortness of breath…), susceptibility to infections, and pain crises caused by poor blood circulation and insufficient oxygenation of tissues (especially the bones). The manifestations are highly variable from one person to another and, for the same person, from one moment to another. If left untreated, patients may develop vaso-occlusive crises, which are due to a poor blood supply to certain organs, and can lead to an "infarction" in the abdomen, chest or skeleton. Sickle cell anemia is one of the diseases that are **systematically screened for at birth** because the consequences will be less serious if it is diagnosed, and therefore treated, early. Management must include, from birth, the prevention of infections and pain, as well as anticipating the need for blood transfusions during crises, which remains an essential treatment. Hydroxyurea may be prescribed for severe forms of the disease. **Clinical trials** are underway.



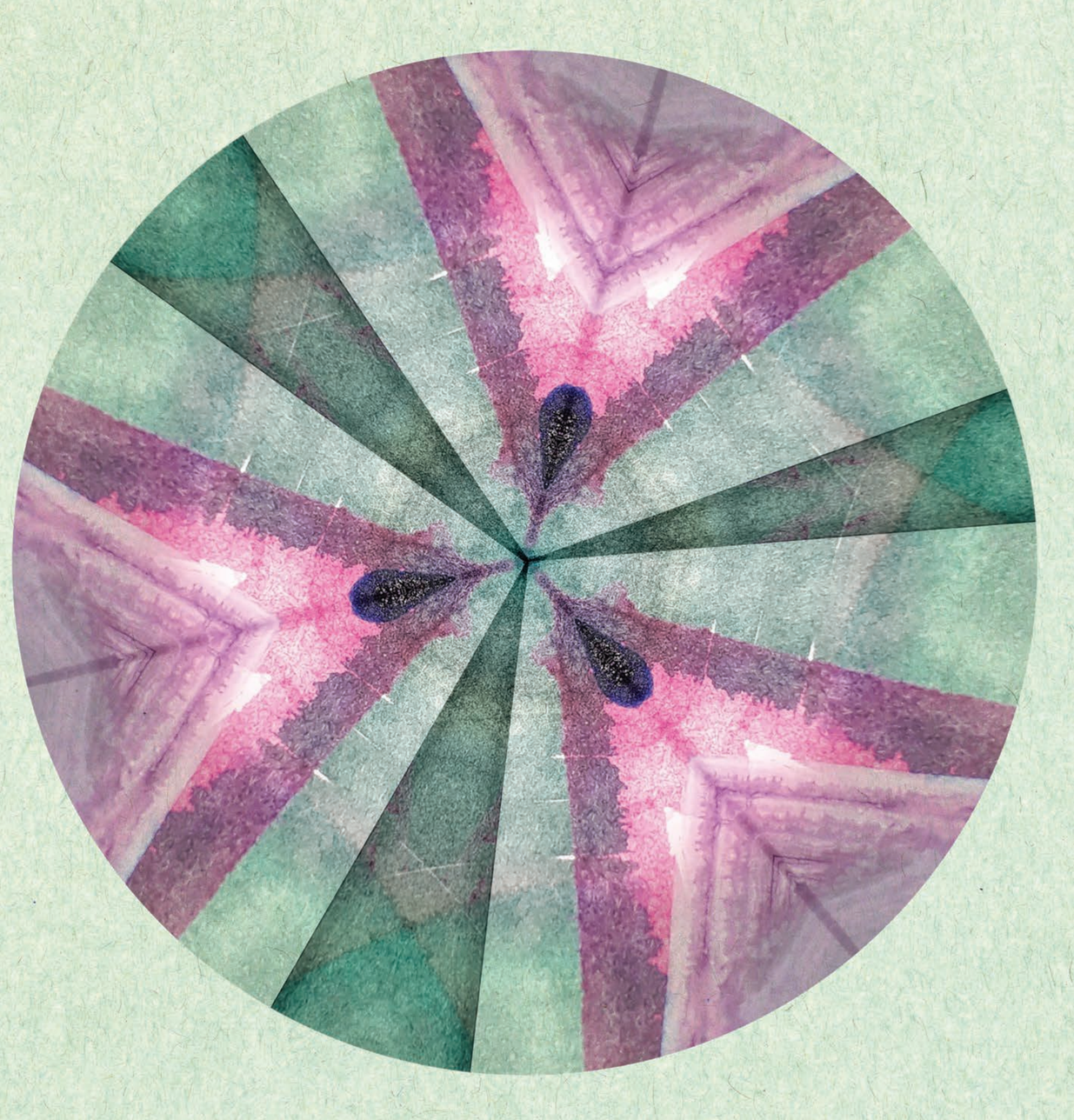


THAUVIN-ROBINET-FAIVRE

Thauvin-Robinet-Faivre syndrome, also known as **tall stature-intellectual disability-renal anomalies syndrome**, is an ultra-rare genetic disorder characterized by **generalized overgrowth**, involving mainly height, and mildly delayed psychomotor development with **learning difficulties** ranging from mild to severe. Less common features may include congenital **cardiac anomalies**, **renal abnormalities**, and **skeletal malformations**. Patients may have an increased risk of developing a renal embryonic tumor (Wilms tumor). To date, only **two families** have been reported in the world, both involving consanguineous parents. Given the ultra-rare nature of the disease, no therapeutic projects can be conducted.

USHER

Usher syndrome is a genetic disease, characterized by the association of progressive hearing loss and **vision disturbances** due to a progressive damage of the retina (retinitis pigmentosa), leading to visual impairment. Depending on the type of Usher syndrome, hearing loss may appear before vision problems or vice versa. Deafness may be present from birth or it may develop later. Vision loss begins in the second or third decade of life. The disease is passed on to offspring when **both parents** carry a variant in one of the causative genes.
Research studies are aimed at identifying **therapeutic strategies** to limit the progression of retinal damage. **Cochlear implants**, or **hearing aids**, can be proposed depending on the degree of deafness, and are most effective when proposed early.



VACTERL

VACTERL/ VATER association is a condition involving multiple **congenital malformations**, characterized by the presence of at least three of the following malformations: vertebral defects, imperforate anus, abnormal tracheo-esophageal connection, renal malformations and limb anomalies. **No intellectual disability** is reported and, in general, patients have a relatively good quality of life. Some malformations are usually detected during pregnancy, while others are detected at birth. The **cause is unknown** and typically there are no familial cases reported. Management is surgical and multidisciplinary.

W

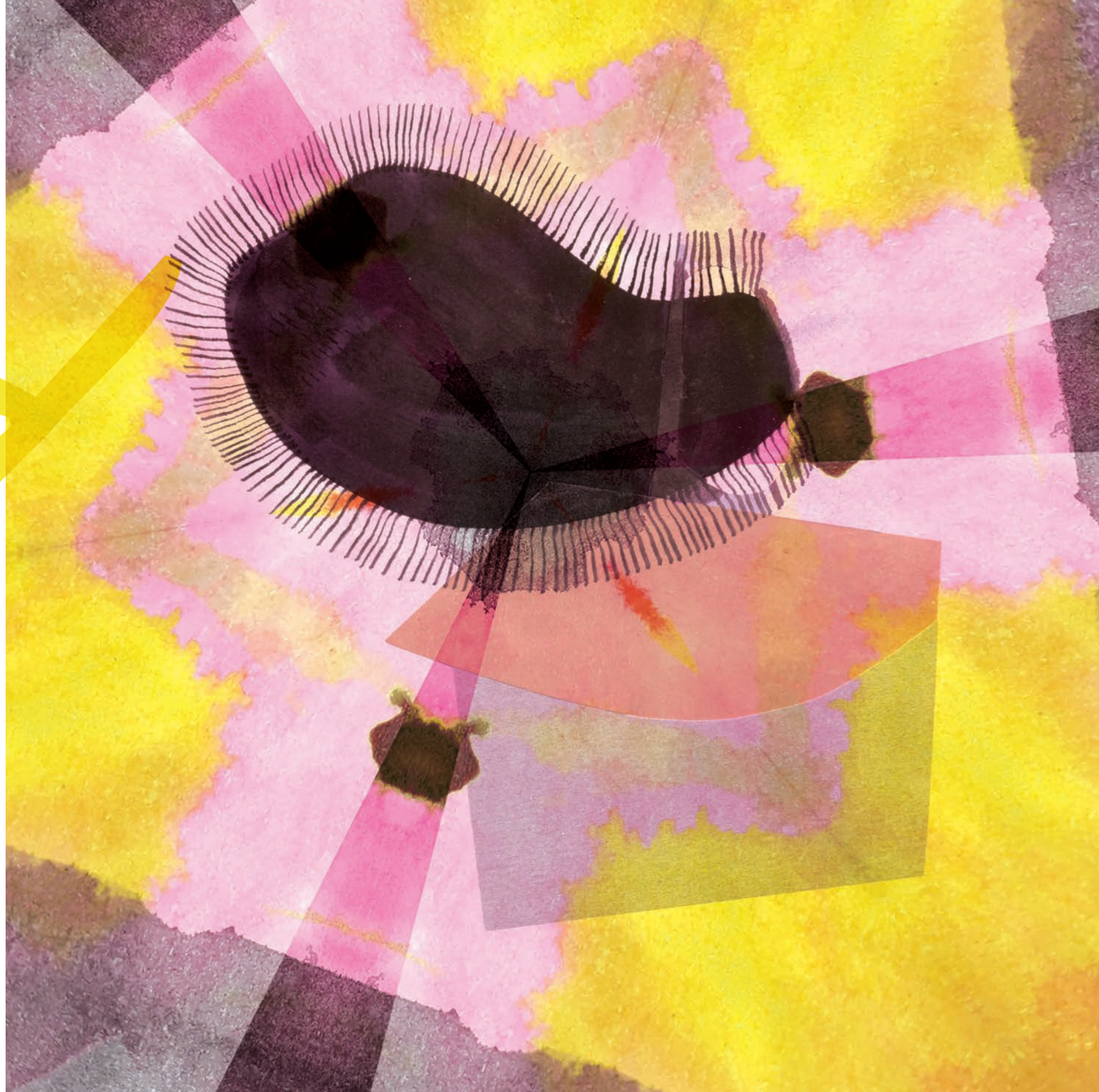
WHITE-SUTTON

White-Sutton syndrome is a rare neurodevelopmental disorder caused by a spontaneous mutation in the *POGZ* gene. It is characterized primarily by **developmental delay**, with or without intellectual disability, **autism spectrum disorder (ASD)**, speech and language delays, and impaired motor skills. The diagnosis is often made in childhood when neurodevelopmental disorders are detected, thanks to new genetic technologies. Treatment is based on rehabilitation and an **individualized education plan**, according to the child's needs. The prognosis depends on the severity of neurodevelopmental problems.

X

XERODERMA PIGMENTOSUM

Xeroderma pigmentosum (XP) is a rare inherited genetic disorder that causes an extreme sensitivity to UV (ultraviolet) light. It is passed on to offspring when **both parents** are carriers of a variant in the gene responsible for the disease. The diagnosis is usually made in infancy. If not fully protected from sunlight, patients experience **accelerated skin aging** and inevitably develop **eye and skin damage** that can lead to multiple types of **cancer**.



my

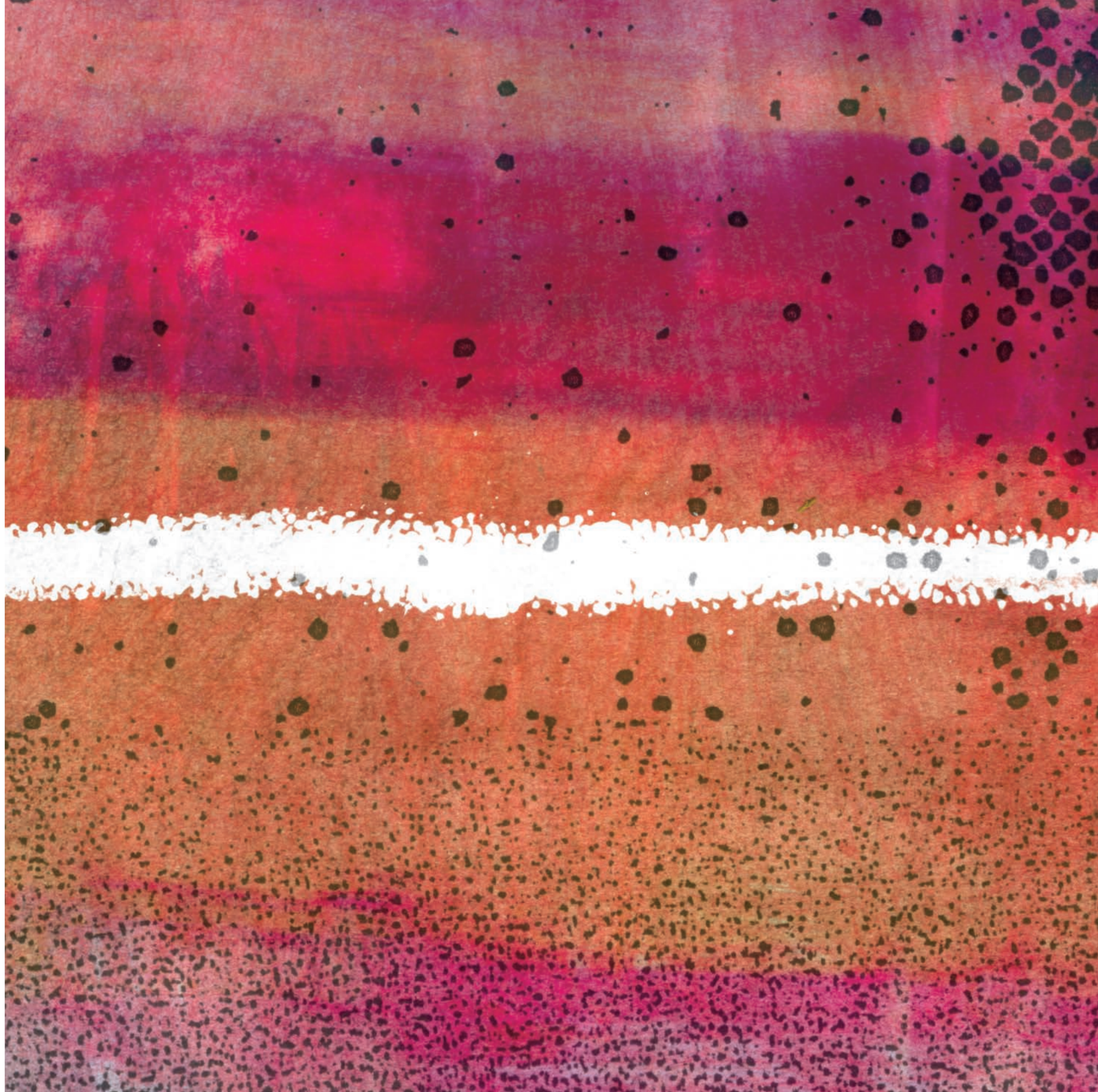
YUNIS-VARON

Yunis-Varon syndrome is a malformation syndrome characterized by **skeletal anomalies of the skull and clavicles**, characteristic facial features, **bilateral absence of the thumb** and first metatarsal, and **absence of distal phalanges**. Brain malformations are often associated. Abnormalities are usually detected during pregnancy, in which case termination of pregnancy may be considered. From birth, affected individuals present with **decreased muscle tone** and overall **developmental delay**, as well as **breathing, feeding and swallowing difficulties**.
In most cases, death occurs within a few months after birth. It is an ultra-rare syndrome, which is passed on to offspring when **both parents** carry a defect in the gene responsible for the disease.
No treatment exists to change the course of the disease.

Z

ZELLWEGER

Zellweger syndrome is a congenital (meaning present at birth) disease characterized by the **reduction, or absence, of peroxisomes** (cellular structures that rid the body of toxic substances) in the cells of the liver, kidneys and brain. This disease manifests with very **characteristic facial features**, severe hypotonia (muscle weakness), **epileptic seizures** and l**iver and kidney dysfunction**. The diagnosis is usually made in the first few weeks of life. Affected children often die within the first year of life. It is a rare syndrome, which is passed on to offspring when **both parents** are carriers of an alteration in the gene responsible for the disease. There is no specific treatment, except for the management of symptoms and palliative care.



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About the organization of rare diseases in France

More than **8000 rare diseases have been recorded worldwide**. According to the European definition, a disease is considered "rare" when it affects less than one in 2000 people in the general population. Therefore, a rare disease can affect anywhere from a few people to several thousand. **They affect between 3 to 4 million people in France** and more than 30 million people in Europe.

Rare diseases are therefore an important public health issue. They concern all medical specialties and their severity varies greatly depending on the disease and the individual patient. Numerous and complex, they are often **poorly known by the medical community**.

Indeed, due to their low prevalence, these rare diseases are misdiagnosed or undiagnosed, and are managed very unevenly between medical institutions.

For the majority of these diseases, there is no curative treatment.

80% of rare diseases are of genetic origin. The remaining 20% correspond to very rare infectious diseases, autoimmune diseases or rare cancers.

For most, the cause remains unknown to this day. We speak of **diagnostic wandering or diagnostic odyssey**.

Given the importance that early care can have in improving the quality of life and survival of patients, the need to organize expert centers specialized in the management and treatment of rare diseases became apparent.

In France, three national plans dedicated to structuring the management of rare diseases have been successively implemented.

2005-2008

RARE DISEASE **EXPERT CENTERS**

The creation of rare disease expert centers, accredited for 5 years, was one of the missions set out in the **1st National Plan for Rare Diseases (PNMR 1 2005-2008)**:

- The **Rare Disease Reference Centers (CRMR in French)** are reference centers recognized for their expertise in the management and care of people with rare diseases and for their commitment to research and training. The cystic fibrosis, rare hemorrhagic diseases and ALS **Resource and Competence Centers (CRC in French)** joined the CRMR network.
- The **Rare Disease Competence Centers (CCMR in French)** ensure the care and follow-up of people with rare diseases as close as possible to their homes, based on an adapted territorial network and in conjunction with the CRMR on which it depends functionally.

2011-2016

THE FRENCH RARE DISEASE **NETWORKS**

The **2nd National Plan (PNMR2 2011-2016)** continued in the same direction by developing **23 French Rare Disease Networks (FSMR in French)**. An FSMR is an organization that coordinates a network of CRMRs, CCMRs, CRCs, healthcare professionals, diagnostic and research laboratories, educational, social and medico-social structures, universities, patient organizations and any other partner - including those from the private sector - that provides added value to the collective action.

The PNMR2 equally promoted the establishment of the **European Reference Networks (ERNs)**. These are networks of centers of expertise and health care providers organized on a European scale in order to allow clinicians and researchers to share knowledge and resources.

2018-2022

THE **PLATFORMS OF EXPERTISE** FOR RARE DISEASES

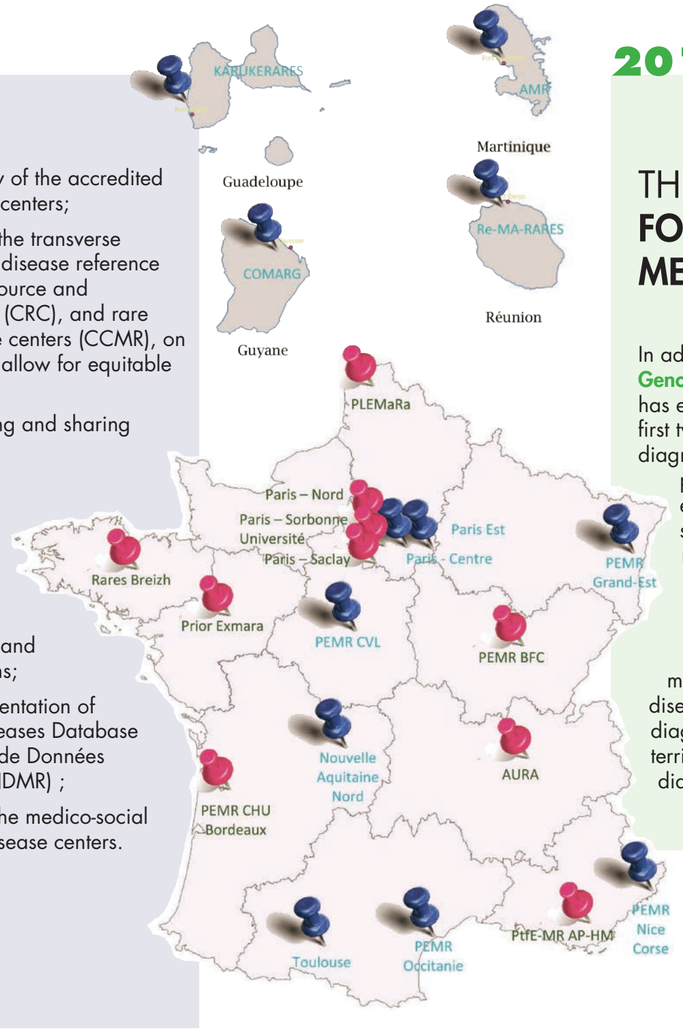
The **3rd National Plan (PNMR3 2018-2022)** is articulated around 5 ambitions:

- research and innovation ;
- quicker diagnosis in order to reduce diagnostic wandering;
- improving the quality of life of patients;
- communication and training ;
- modernization of organization procedures and funding optimization.

The **Platforms of Expertise for Rare Diseases (PEMR in French)** are part of the actions set up within the framework of the PNMR3. These PEMR aim to strengthen inter-network links within the host establishments of several accredited reference centers.

The purpose of these platforms is to **share expertise and pool knowledge, expertise and resources at a local level** in order to:

- improve the visibility of the accredited rare disease expert centers;
- share resources for the transverse missions of the rare disease reference centers (CRMR), resource and competence centers (CRC), and rare disease competence centers (CCMR), on a territorial scale to allow for equitable distribution;
- participate in training and sharing of information;
- support diagnostic and therapeutic innovation, as well as research;
- strengthen the links between centers of expertise and patient organizations;
- promote the implementation of the French Rare Diseases Database (Banque Nationale de Données Maladies Rares; BNDMR) ;
- facilitate links with the medico-social actors in the rare disease centers.



2018-2025

THE FRENCH **PLAN FOR GENOMIC MEDICINE**

In addition, the **French Plan for Genomic Medicine (PFMG) 2025** has enabled the establishment of the first two genomic laboratories for diagnostic and therapeutic monitoring purposes. These facilities of excellence illustrate the ongoing support of the public authorities for medical innovation, in this case the very high-speed sequencing of the human genome, which is the basis of genomic medicine, also known as "personalized" medicine. In the field of rare diseases, they aim to reduce diagnostic delays, and to allow for territorial equity in terms of access to diagnosis.

The plan has 3 objectives:

- to prepare for the integration of genomic medicine into the current care pathway and the management of diseases. The goal is to guarantee access to genomic medicine for patients who need it, whether they have cancer, a rare disease or, in the long term, a common disease;
- to set up a national genomic medicine network for the benefit of patients, capable of being a catalyst for scientific and technological innovation, industrial development and economic growth;
- to place France at the forefront of the major countries involved in personalized medicine, with a capacity to export the know-how of our medical and industrial sector in genomic medicine.

About Lucie Albon

Pencils, a few pieces of paper cut-outs and a little ink… With these components, Lucie Albon is able to invent any universe without ever compromising her artistic originality.

Winner of the Villa Kujoyama, she is equally comfortable working on large scale city murals as she is illustrations for books. She has published more than thirty books for young people at Glénat, L'Élan Vert, Bluedot, Utopique or Rouergue. She has collaborated with Elle magazine and continues to produce artwork for the press.

Trained at the Beaux-Arts d'Angoulême and then at the Arts Décoratifs de Strasbourg, she possesses a great mastery of traditional techniques and the ability to reclaim them in order to achieve a bare simplicity that opens the gates to the imagination…

Website: **www.luciealbon.net**
Agent: **www.patricia-lucas.com**

About the Fondation Ipsen BookLab

At the service of the general interest, working towards an equitable society, the Fondation Ipsen BookLab publishes and distributes books free of charge, primarily to schools and associations. Through collaborations between experts, artists, authors and children, our publications, for all ages and in a variety of languages, focus on the education and awareness of issues related to health, disability and rare diseases. Discover our complete catalog online at **www.fondation-ipsen.org/book-lab**.

About the Platform of Expertise for Rare Diseases – Bourgogne-Franche-Comté (PEMR BFC)

The PEMR BFC is one of the first PEMRs set up in France. The purpose of these platforms is to share expertise and to pool knowledge, skills and resources among different networks and accredited reference centers. This platform is coordinated by the Dijon Bourgogne University Hospital in partnership with the Besançon University Hospital. The two institutions conduct numerous shared projects in the field of rare diseases. The PEMR BFC federates 11 Rare Disease Reference Centers (CRMR) and Resource and Competence Centers (CRC) and 101 Rare Disease Competence Centers (CCMR) belonging to 22 French Rare Disease Networks. The main objectives of the platform are to inform as many people as possible about rare diseases, to enable patients and their families to be directed to expert centers and to support them in their actions, to optimize the care paths in order to adapt them to each individual, to train professionals and to support clinical trials and research in the field of human and social sciences.

Website: **www.pemr-bfc.fr**

Book# 00.4
Have your say!

Texts: Prof. Laurence Faivre, *coordinator*, Elodie Gautier, *project manager*, Sonia Goerger, *administrative assistant*,
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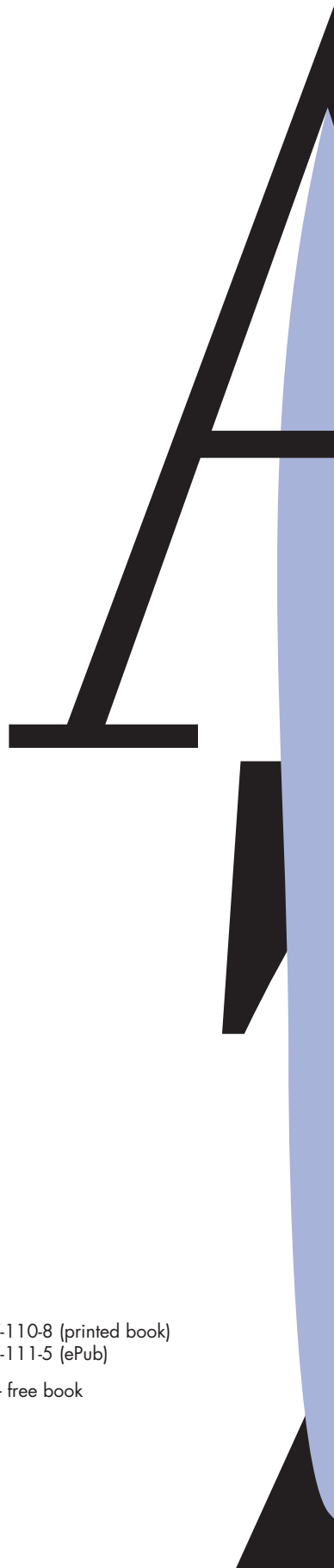
Not for sale – free book

A true sensory experience, this book invites us to discover the diversity and variability of rare diseases. Using a mixture of different visual art techniques, Lucie Albon's artwork conveys the full complexity of rare diseases. As each rare disease has a unique impact on each patient, the images created by the artist speak to the viewer in their own unique way. Yet, in the same way that the 26 letters making up the alphabet form the basis of each word, the rare diseases form a community so that no one should ever feel alone.



Rare Diseases from A to Z, imagined by Lucie Albon, is published in collaboration with the Platform of Expertise for Rare Diseases – Bourgogne-Franche-Comté (PEMR BFC).

PEMR Bourgogne-Franche-Comté Plateforme Expertise Maladies Rares

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