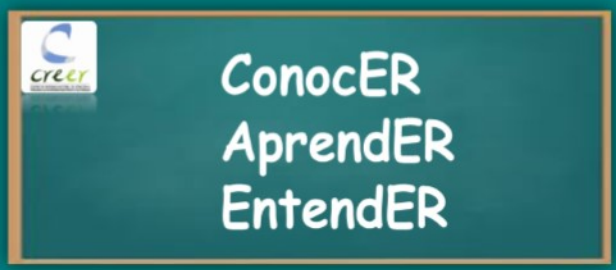
Enfermedad de Dravet Síndrome de Noonan Neurofibromatosis Síndrome de Prader-Willi Síndrome de Sotos Enfermedad de Wilson Síndrome Phelan-McDermid Paraparesia Espástica Familiar Enfermedad de Gileneau Esclerosis Tuberosa Síndrome de West Síndrome de Angelman Síndrome de Lenox-Gasteau Hipotiroidismo Congénito Déficit Congénito de Glicosilación Galactosemia Fenilcetonuria Enfermedad de Huntington Fiebre Mediterránea Familiar Síndrome de Joubert Síndrome de Poland Síndrome de Moebius Displasia Ectodérmica Epidermólisis Bullosa Enfermedad de Gaucher Fibrosis Quística Síndrome de Lesch-Nyhan Leucodistrofia Enfermedad de Pompe Albinismo

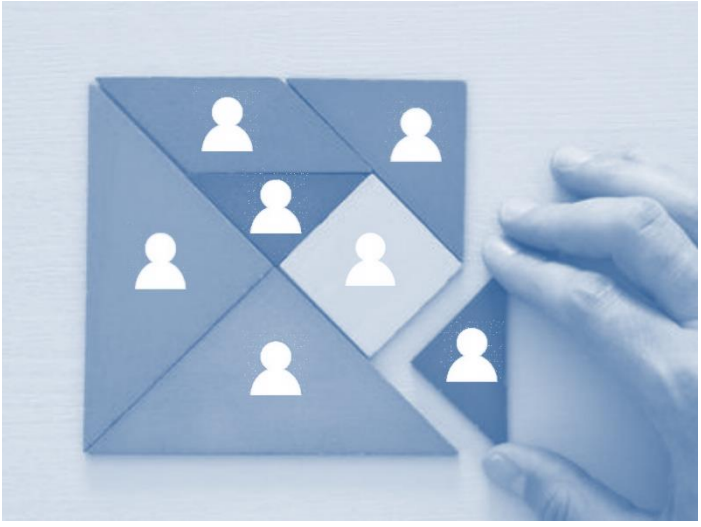
RARE DISEASES IN SCHOOLS

support guide



GOBIERNO DE ESPAÑA
MINISTERIO DE DERECHOS SOCIALES Y AGENDA 2030
SECRETARÍA DE ESTADO DE DERECHOS SOCIALES
IMSERSO
AGENDA 2030

RARE DISEASES
IN
SCHOOLS



SUPPORT GUIDE

PROMOTED BY:

National Reference Centre for People with Rare Diseases and their Families (Creer), belonging to Institute for Older Persons and Social Services (Imserso)

PUBLISHED BY:

National Reference Centre for People with Rare Diseases and their Families (Creer), belonging to Institute for Older Persons and Social Services (Imserso)

AUTHORS:

Yolanda Ahedo Infante

Montserrat Cabrejas del Campo

Ana Santamaría Herrera

TRANSLATED BY:

Centro de Referencia Estatal de Atención a Personas con Enfermedades Raras y sus Familias (Creer), belonging to Instituto de Mayores y Servicios Sociales (Imserso)

*NIPO: 131200225 (paper spanish edition)

NIPO: 131200230 (pdf spanish edition)

Nº edición: 1ª

* Identification Number of Official Publications in Spain

Dedication

This Guide is especially dedicated to all children and families living and living with a rare disease.

Acknowledgements

We would like to thank the families with rare diseases for the trust they have placed in us since the creation of the National Reference Centre for people with rare diseases and their families (Creer). For all that they have taught us and for the opportunity they have given us to learn about their realities, their experiences, for sharing their dreams, their needs and celebrate their achievements.

Without them, we would not have been able to produce this guide.

We would also like to thank the schools that have faithfully welcomed us and support us in the task of making visible and bring the reality of the rare diseases closer to the school.

Yolanda Ahedo Infante
Montserrat Cabrejas del Campo
Ana Santamaría Herrera

Creer's Education Department

The **National Reference Centre for People with Rare Diseases and their Families (Creer)**, is a public resource of state level, created and regulated by the order SAS 2007/2009 of the Ministry of Health, Social Policy and Equality with the strategic aim of promoting innovation and improve the quality of resources for people with rare disease and improve their quality of life.

It was inaugurated in 2009 and it is oriented to the promotion of resources, services, equipment, methods and techniques for intervention in the whole territory of the State. It is framed in the Imserso, Management Entity of the Social Social Security, currently under the Ministry of Social Rights and 2030 Agenda.

The Center has a multidisciplinary team of social, health and educational fields.

CREER'S EDUCATION DEPARTMENT

Objective:

Improve the quality of life of students with Rare Diseases and their families, by supporting and favoring their educational inclusion.

Areas of Intervention:

Individual and/or group attention for minors and their families.

Information, counselling and coordination with counselors, tutors,… of the schools.

Awareness Raising and Dissemination Program in schools.

Specialized Training of professionals, in active and training, in the field of Education.

Participation and collaboration in projects with other centers or entities.

TABLE OF CONTENTS

1. PRESENTATION OF THE GUIDE

Little is known about **Rare Diseases (RD)** at health, social and educational level. Different studies highlight the general lack of information within the educational community amongst teachers, carers, classmates etc.

This guide to Rare Diseases is written for the educational community and aims to provide information about RD in general and offer support, resources, materials, strategies, etc. in a bid to improve the quality of life of pupils with an uncommon disease.

There is no such thing as a magic solution, particularly in education where each student is different and has his/her own specific needs. But we do believe that knowledge and information can open doors and help us to **Get to know, Learn and Understand about RD**.

This is what we hope to achieve with this guide, to open spaces for reflection, imagination, flexibility and in short to approach this issue from an alternative perspective so as to be able to create more **inclusive** schools.

We do not aspire to turn you into experts in RD but we would like you to have a general vision that would enable progress in three main areas:

- To better understand the needs of these students.
- Encourage the development of tolerance, respect and acceptance of diversity in the classroom.
- Raise awareness about RD so as to improve relations between students.

If this guide falls into your hands, please devote a few minutes of your time to it

2. INTRODUCTION

Students with rare diseases are a very diverse group. When we talk about rare diseases, we are faced with a wide range of disorders and symptoms that vary, not only from one disease to another, but also within the same disease.

"Health conditions which, in the sense that they are varied and diverse, affect the lives of the people concerned and their families in the same way and to the same degree, in terms of both their needs and of the response they require from such varied fields as health, education, social services, etc., in order to guarantee their quality of life" (Javier Monzón, 2014).

The day-to-day lives and the needs of pupils with uncommon diseases are complex and affect a whole array of different aspects of the teaching and learning process.

Inclusive Education entails a dialectic process between presence, learning and participation. **Presence**, to find each other in common spaces. **Participation**, to enable us to be recognized for what we are, to be accepted, valued and RESPECTED. And **Learning**, understood as achieving the maximum possible development of the capacities of EACH AND EVERY ONE OF THE PUPILS, including the boys and girls with Rare Diseases.

- ✓ **We hope that this guide will be useful for consolidating inclusive education.**

3. CONCEPTUALIZATION

3.1 Why are they called Rare Diseases?

A disease is considered rare or uncommon when it affects no more than one in every 2,000 people.

This is the agreed prevalence figure in the European Union.

Lymphangioleiomyomatosis **Ana**
Trimethylaminuria **Luis**
Hirschsprung **Teresa**

The Spanish name "*enfermedades raras*" is a literal translation of the English term "*rare diseases*". In English "rare" is used to describe something scarce or in very short supply. In Spanish, however, the word "raro" has pejorative connotations, referring to things that are "weird", "odd" or "strange". Some Patients Associations therefore support the use of other terminology.

SYNONYMS: infrequent diseases, minority diseases, uncommon diseases, orphan diseases

**Prevalence: In epidemiology, the number of people who suffer from any disease as a proportion of the total of the study population.*

3.2 How many Rare Diseases exist?

So far, over 7,000 different minority diseases have been identified.

In Spain, an estimated **three million** people have to **live with an RD.**

Three million = patients, family, professionals, carers, society in general.

Puzzle design made up of group photographs of different rare disease associations.

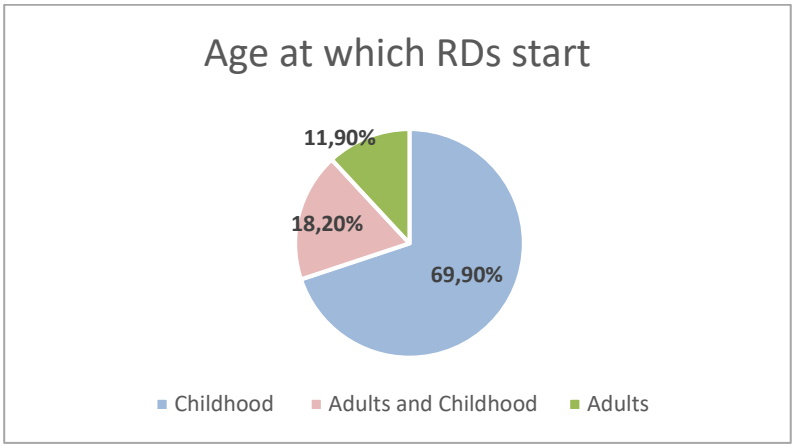
3.3 What are the causes?

80% of infrequent diseases have been identified as of **genetic** origin.

20% are due to **other causes** (infections, allergies, teratogenic, proliferative, unknown).

3.4 Who might they affect?

Due to the high percentage of diseases of genetic origin and of congenital anomalies, most minority diseases appear in childhood, although they can appear at any time of life.



Graph: Created by the authors with data from an article entitled "Estimation of the accumulated point prevalence of RD: analysis of the Orphanet database" (2020).

69.9% appear exclusively during childhood
18.2% can start in both childhood and adulthood
11.9% occur exclusively in adults

✓ **The fact that cases appear during childhood means that their symptoms and characteristics are present throughout the child's school and academic career**

4. CHARACTERISTICS OF RARE DISEASES

The over 7,000 infrequent diseases described so far share the following characteristics:

- **Difficult diagnosis**: the average delay in diagnosis is 5 years
- **Lack of curative treatments**: many only treat the associated symptoms
- **Chronic nature**: the disease accompanies those affected throughout their lives
- **Progressive and degenerative evolution**: gradual loss of abilities and autonomy
- **Multisystemic affection**: they can cause physical, sensorial, mental, intellectual and organ disability
- **Non-visible effects:** pain, tiredness, fatigue, anxiety, low self-esteem, fear…
- **Lack of information**: due to their low prevalence
- **Need for a comprehensive programme of care**: they require coordinated care from different professionals
- **Little research**: due to their high costs, low returns and the limited number of case studies…

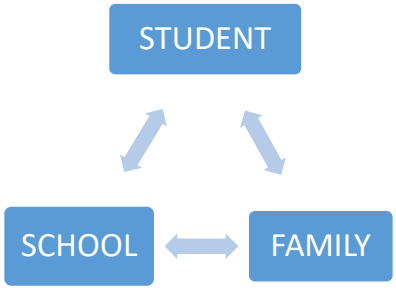
5. CONSEQUENCES OF HAVING A RARE DISEASE

Rare diseases, independently of their associated clinical issues and on many occasions exacerbated by them, have consequences for all aspects of life, at a personal level and in our family, educational, social and working lives and on all the different planes - physical, emotional, psychological, cognitive etc.

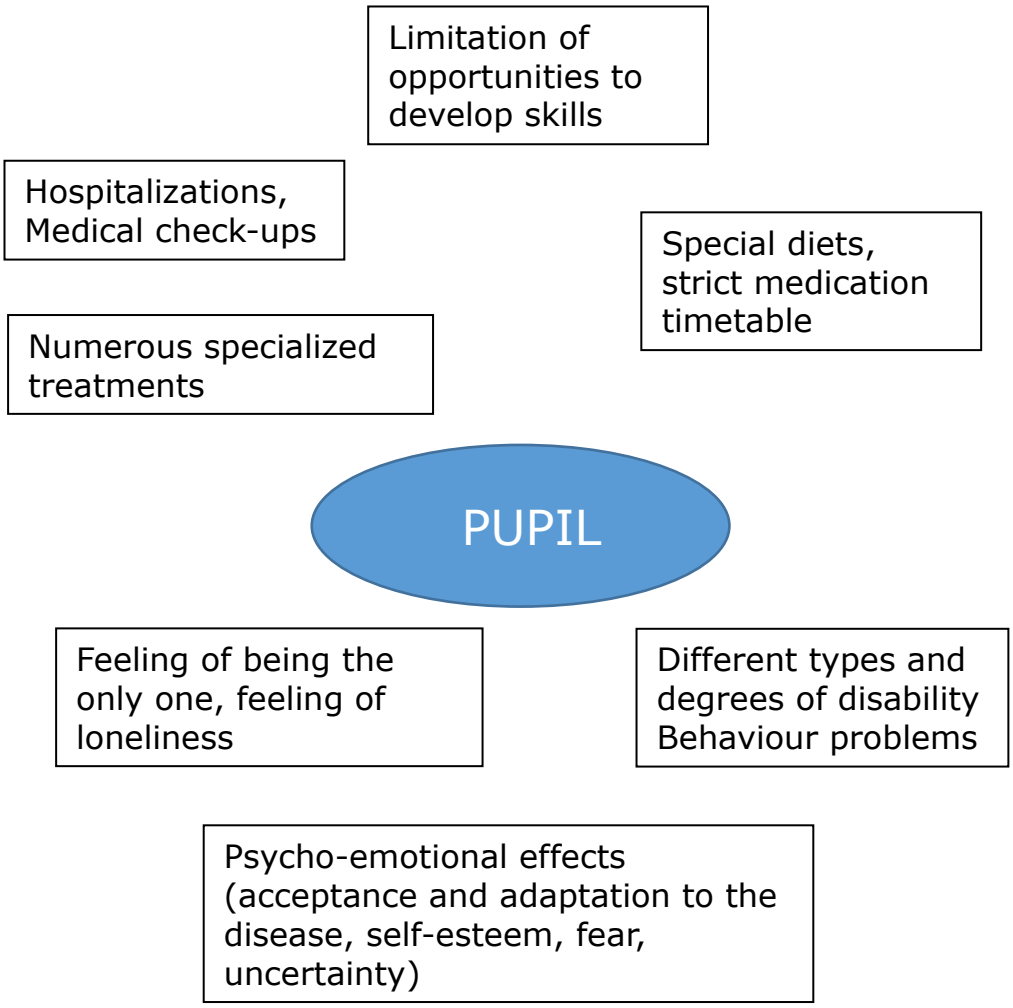
These consequences of rare diseases impact not only on the person directly affected but also on their family, school and social environment.

RD damage the quality of life of the person concerned, their family and wider circle, which is why a better knowledge of these diseases will enable us to make adjustments that can help improve their quality of life.

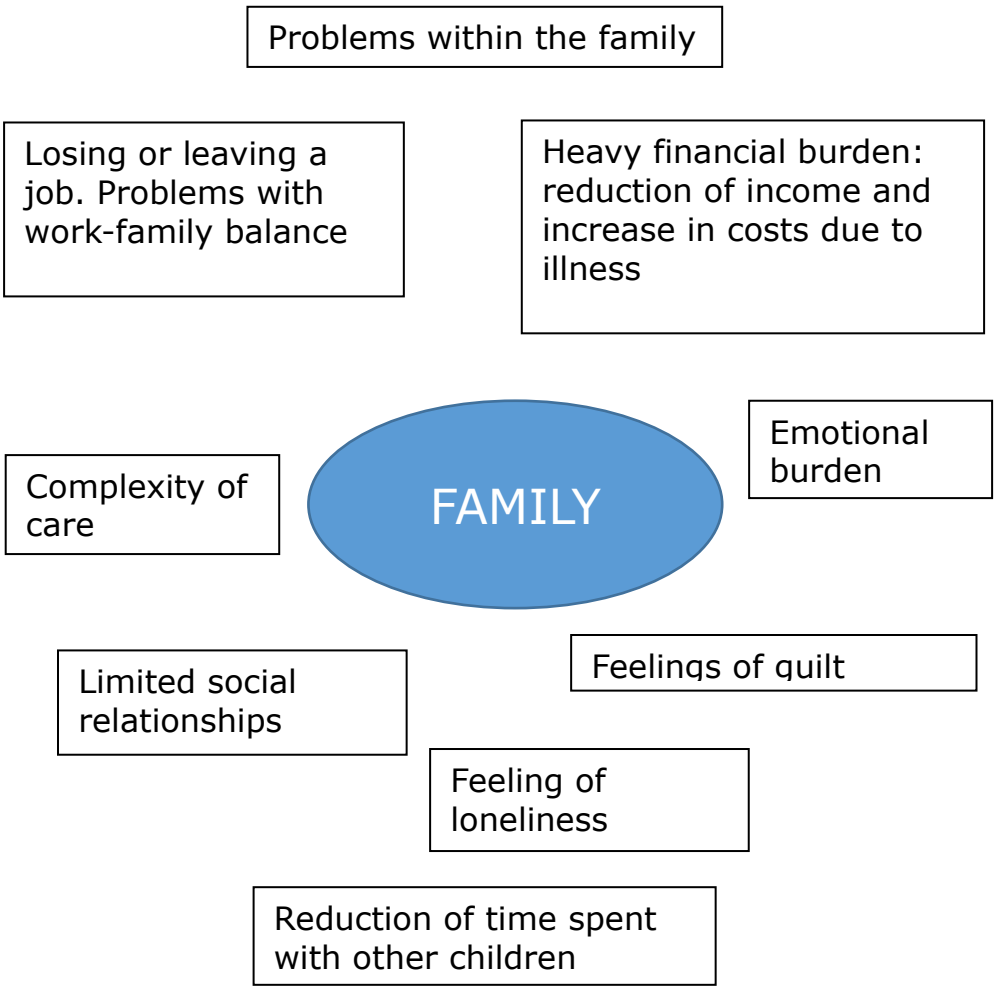
For children with rare diseases, being able to go to school plays an essential role in the normalisation of their everyday lives, and so becomes a factor in their health.



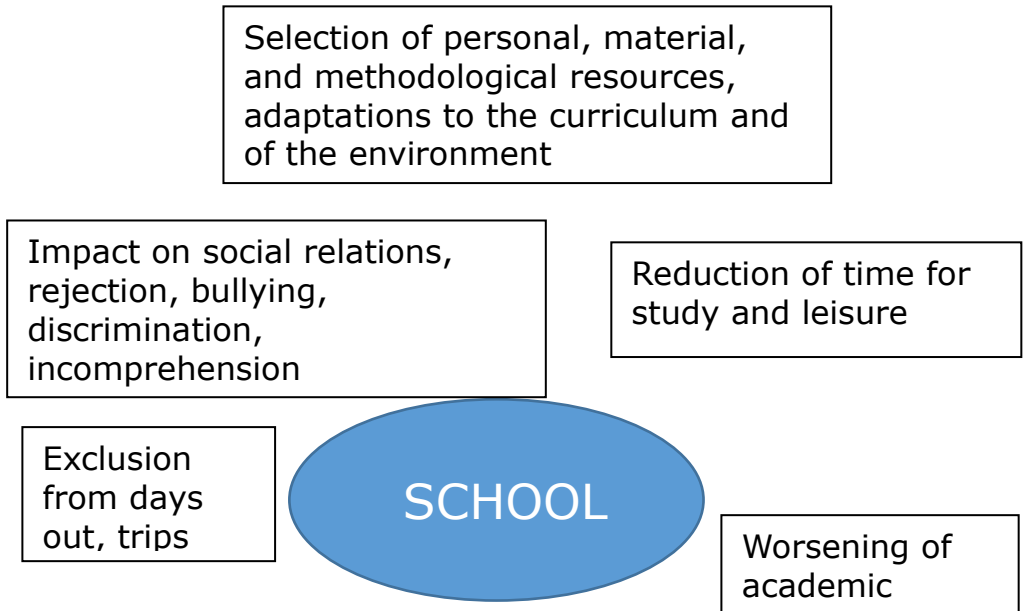
✓ **The consequences of RD impact not only on the person directly affected but also on their family, school and social environment**



- ✓ **These consequences vary depending on the patient's medical history and age.**
- ✓ **All of this makes pupils with an RD more vulnerable and at greater risk of failing at school.**



- ✓ **These problems can cause the families to shut themselves away, so making family-school communication more difficult**



✓ **The right adaptations to deal with these problems will enhance educational inclusion and help guarantee quality of life.**

6. WHAT CAN WE DO AS A SCHOOL TO IMPROVE THE INCLUSION OF PUPILS WITH RD?

Just like all other pupils, those with RD have a wide array of needs, the special thing in their case being that their diseases are **less well known** in the educational field and **can go unnoticed**.

Before deciding what measures to take, it is necessary to **get to know, learn and understand** the everyday reality and characteristics of rare diseases.

It is crucial to correctly **identify the educational needs** (not always immediately visible) of students with rare diseases.

- ✓ **The needs of students with RD may not be noticed at school.**

Educational inclusion of students with minority diseases must be based on three basic pillars

A. WARM WELCOME
B. IDENTIFICATION OF NEEDS
C. ADAPTED RESPONSE

A. A WARM WELCOME

In the first contact with the family, it is important to create a special atmosphere of confidence. Many of them, due to the experiences they have had throughout their lives, (the search for a diagnosis, the lack of information, the emotional load, etc.) are going to need and will be grateful for a warm welcome full of empathy, acceptance and support.

Not all rare health conditions are associated with special educational needs, but they can impose limitations and conditions, due to their clinical effects, treatments, progression or due to the lack of diagnosis, or the personal, social and academic adaptation of the student.

Bearing all this in mind and the consequences, we referred to earlier, in the **Warm Welcome** we recommend:

- **Gathering** information about all aspects of their lives: health, skills, abilities, expectations and their consequences for the teaching-learning process.
- **Coordinating** with other professionals from different systems that care for or attend to the child: health, social and educational workers.
- **Looking for reliable documentation** about the disease and its evolution.
- **Finding out and respecting** the opinions of both the pupil and their family, accepting their right not to inform others. The diagnosis of a rare disease is sometimes accompanied by a fear of the label, prejudice, rejection etc.

How do we obtain the information we need for the welcome?

Reliable information can best be obtained through the family, in coordination with professionals who have attended to them in the past, through public or private services specializing in the care of people with rare diseases.

✓ A warm **Welcome** lets us **Get to know**

B. IDENTIFYING NEEDS

It is important to bear in mind that RDs may appear at any time in a child's school career and are not always present at the beginning of the school year. Special sensitivity is required to be able to identify them.

In addition, the progressive nature of these illnesses will lead to changing, cumulative needs, even within the same school year.

✓ **Identifying the needs of students with a minority disease requires continuous observation.**

What needs do pupils with RD have?

- Those set out in the pupil's Psycho-Pedagogical Report, if there is one.
- Those identified through regular, fluid communication with the family.
- Those specified in the pupil's medical reports.
- Others that may manifest themselves at school which are more difficult to identify as they are not so visible, but which may have serious effects on school performance, such as: **tiredness** (many RD upset the wake-sleep rhythm), **physical and emotional pain** (either chronic and permanent or from time to time in diseases that appear in sporadic bouts…), **uncertainty, fear, nervousness, stress**.

The key for these pupils is to find the right balance between academic, emotional and health objectives.

> ✓ Proper **Identification of their educational and health needs** allows us to **Learn**

C. PINPOINTED RESPONSE

In order to respond to the needs identified, the schools can apply measures for attention to diversity.

Measures for attention to diversity*

General ordinary measures aimed at all the students, which do not alter any essential elements of the curriculum and are aimed at methodological and organizational strategies (tutoring action plan, welcome plan, doubling up, ordinary tuition support, planning time, spaces,…).

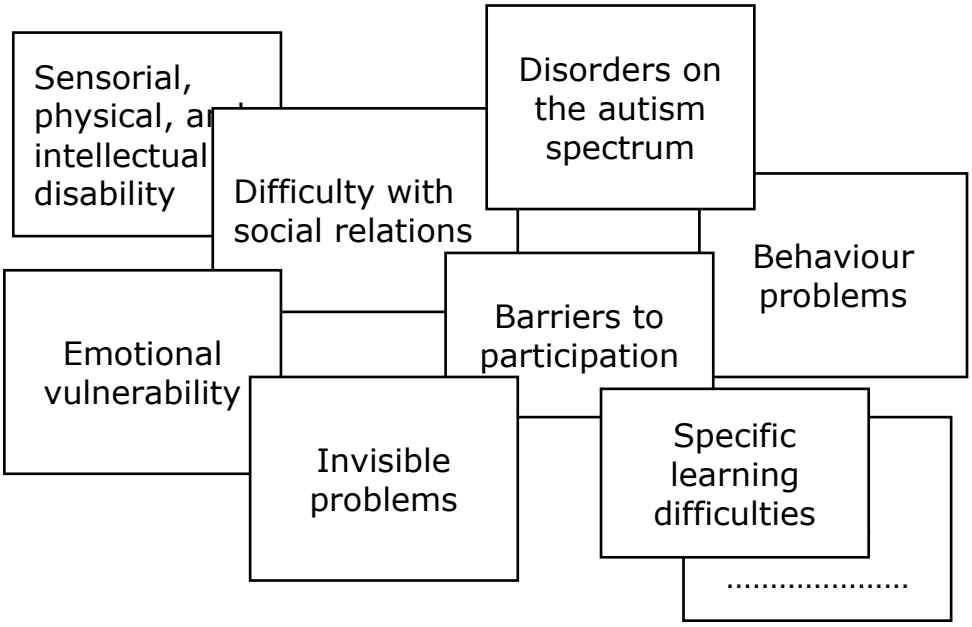
Specific extraordinary measures aimed at a specific profile of pupils, which could change essential aspects of the curriculum such as objectives, contents, assessment criteria (combined or specialized schooling, home care, hospital classrooms, specialized personnel and material resources…).

Exceptional measures that do not change essential aspects of the curriculum, whose objective is to enable RD pupils to succeed at school (extending each stage of their school career by one year, being flexible about the period that each student must spend on each stage, acceleration).

***** *The name we use in this guide seeks to unify the different terms used to refer to this concept in the different Spanish regions.*

- ✓ **Pupils with an uncommon disease are typically quite diverse, with educational needs of different kinds, degrees and levels**

Within this diversity, we may find:



Attention to diversity measures tailored to pupils with RD

A tailor's measuring tape

- Support academic continuity in cases of repeated absence for health reasons, by providing online, digital tools that enable the pupils to continue with their schooling and remain connected to school life.

- Normalize the stigma associated with "rarity" or "oddness", always with the permission of the family and of the pupils themselves, by organizing information and awareness-raising activities aimed at the entire school community.

- Encourage classmates to contact the pupil during their absences through letters, messages, video calls, etc.

- Share relevant information about the pupil, to the extent that it affects his/her learning or physical wellbeing, with all professionals involved (teaching and non-teaching personnel).

- Appoint someone as the "professional of reference", who does not necessarily have to be the class tutor, to channel all information with regard to the pupil (personality, current progress of the disease, style of learning, family situation, social relations …).

- Plan activities, excursions, taking into account all the children at the school, while trying to anticipate the resources and support that may be required to facilitate the participation of a pupil with an uncommon disease.

- Consider collaborating with families, rare disease associations and other entities, as a source of reliable information that will help ensure a satisfactory school experience.

- Organize work meetings with professionals from the health field to find out about activities/programmes, special care, how to deal with emergencies, etc.

- Organize and plan classroom activities, homework, assessments, etc. in a flexible way, bearing in mind the effects of the medication, the additional burden of external therapies, clinical issues and the different treatments associated with the disease.

✓ **The measures that facilitate the presence, participation and success of these pupils encompass all the spaces, services and activities of school life. We must therefore also bear in mind the special needs that the pupil may have, so as to enable them to use school transport, catering facilities, early riser programmes, after-school activities.**

✓ We can only **Respond** correctly when we are able to **Understand**

7. INSPIRING INITIATIVES

Progress is being made, albeit slowly, in furthering our knowledge of pupils with RD and there is an increasing number of initiatives and studies that provide insights into this minority group and offer us models for action that can guide our decisions in relation to their schooling.

✓ 7.1 Protocols

Protocol for welcoming and attending to children with rare diseases in schools in Extremadura.

The Department of Education and Employment and the Department of Health and Social Policy of the Regional Government of Extremadura have drawn up this protocol with the participation of the Delegation in Extremadura of the Spanish Rare Diseases Federation (Feder).

The objective of this protocol is to make it easier to incorporate pupils with rare diseases into the school and to implement the necessary measures to provide a suitable health and educational response that meets the needs of these pupils within the school environment.
https://www.educarex.es/pub/cont/com/0004/documentos/PROTOCOLO_ENFERMEDADES.pdf

Provincial Plan for Attention to pupils with chronic diseases

Protocol for coordinated action to improve the attention provided to pupils from a health, organizational and educational perspective.
https://equipotecnicoorientaciongranada.files.wordpress.com/2014/09/plan-de-atencic3b3n-a-alumnado-con-enfermedades-crc3b3nicas-granada-1.pdf

✓ 7.2 Raising Awareness

The National Centre of Reference for Attention to People with Rare Diseases and their Families (El Centro de Referencia Estatal de Atención a Personas con Enfermedades Raras y sus Familias - Creer), has been carrying out awareness raising and dissemination activities about RD in schools ever since its creation in 2009.

Through these activities, the Centre tries to reach as many children as possible and to raise awareness throughout the educational community as to what it means to live with an RD, and to nurture amongst children values such as respect for diversity, empathy, solidarity and mutual aid, among others. The aim is to help normalize the presence of these pupils in the classroom and bring about a change in attitudes and the greatest possible inclusion of these pupils in schools.
"Programme for Raising Awareness and Disseminating Information about Rare Diseases in Schools".
https://creenfermedadesraras.imserso.es/InterPresent1/groups/imserso/documents/binario/difusioncolescreer.pdf

Other organizations in the Rare Diseases field also carry out awareness raising and dissemination campaigns about Rare Diseases in Schools.

Spanish Rare Diseases Federation (Federación Española de Enfermedades Raras - Feder)
http://enfermedades-raras.org/

Spanish Association of Mastocytosis and related diseases (Asociación Española de Mastocitosis y enfermedades relacionadas)
http://www.mastocitosis.com/proyectos-aedm/super-crom

And many other associations…

- ✓ **It is important to repeat the need to offer information and action guidelines to all members of the educational community, focusing particularly on the peer group.**

A group of children sitting on the floor carrying out an exercise within the Creer awareness raising programme

✓ 7.3 Specific guides

In addition, the commitment and collaboration between the educational administration, the organizations specializing in attention to children with Rare Diseases and the Patients Associations has materialised in **Specific Guides for these pupils**.

The purpose of these guides is to provide information to the educational community about the characteristics of the disease and its effects on the teaching/learning process, and on the general functioning of the school, so as to eliminate the initial uncertainty, fear and rejection regarding the schooling of children with RD and correct the wrong interpretations and "labels" regarding behaviour and the functioning of the school. The Guides also help schools draw up a plan that covers the educational needs and is adapted to the real situation of pupils with a rare disease.

➢ Duchenne Muscular Dystrophy

Duchenne Muscular Dystrophy (DMD) is a neuromuscular disease characterized by rapidly progressive muscle atrophy and weakness as a result of the degeneration of smooth, skeletal and cardiac muscles.

The disease begins in early childhood and affected children can suffer delays in reaching the milestones of motor development or delays in overall development. In general, children affected by DMD are unable to run or jump. The disease progresses quickly and the child develops an atypical gait with hypertrophy of the calves. They begin to have trouble using stairs and start falling frequently. They lose their ability to walk between 6 and 13 years of age. Once they are unable

to walk, they quickly develop joint contractures and scoliosis. In some cases, it can also have effects at cognitive level. Cardiomyopathy and respiratory insufficiency may be a cause of death during adolescence.
https://www.duchenne-spain.org/wp-content/uploads/2019/05/WEB-Duchenne-Spain-GuiaEscolar.pdf

- **Aniridia**

Aniridia is a hereditary eye disease, the most evident symptom of which is the absence of the iris, which results in low levels of vision (no more than 20 %) and is caused by a lack of development of the eyeball during pregnancy. It also has a number of associated abnormalities such as glaucoma, corneal degeneration, nystagmus, congenital cataract and other more serious and disabling disorders such as WAGR Syndrome or Wilms Tumour, which result in a much poorer quality of life for the patients and their families.
https://aniridia.es/wp-content/uploads/2016/03/el-Libro-blanco-de-la-Baja-Visi%C3%B3n-en-la-Educaci%C3%B3n-.pdf

- **Epidermolysis bullosa**

Epidermolysis bullosa is a genetic disease, whose most visible characteristic is the extreme fragility of the people that suffer from it. Their skin is as fragile as the wings of butterflies, which is why it is popularly known as "Butterfly Skin". Everyday actions such as walking or eating may be extremely painful. It requires treatments of between one and four hours each day or every other day with special bandages and

materials that can cover the entire body. In addition, the disease also affects other less visible, but just as important, parts of the body: the mucous membranes, eye tissues, the mouth, the digestive tube, etc. Other manifestations of the disease include: anaemia, syndactyly (when fingers or toes are fused together), dysphagia (difficulty swallowing), malnutrition, constipation, osteoporosis, muscular dystrophy, cardiomyopathy, renal insufficiency and cancer among others. https://d3afc8hatrp5rg.cloudfront.net/wp-content/uploads/2019/03/Escolarizacin-del-nio-con-Piel-de-Mariposa.pdf

- **Haemophilia**

Haemophilia is a genetic disease characterized by spontaneous or prolonged haemorrhaging due to deficiencies in the coagulation factors. It affects above all males, although female carriers of the mutation may also present a minority form of the disease. The seriousness of the clinical manifestations depends on the scale of the coagulation factor deficiency. A characteristic sign of the disease is quite frequent spontaneous hematuria.
http://fedhemo.com/wp-content/uploads/2015/05/Cuadernillo-Hemofilia-para-profesores.pdf?platform=hootsuite

- **Albinism**

Albinism is a genetic condition that produces a very characteristic phenotype (physical appearance), due to the absence or low levels of pigment in the skin, eyes and hair. This is an unusual metabolic condition caused by a defect in the gene responsible for the synthesis and distribution of melanin. It affects all races in the same way. Albino people typically have white or slightly golden hair, very pale or pink-tinged skin, and reddish or violet-blue eyes. We notice this reddish colour as the light strikes their eyes, as the lack of pigment allows us to see the reflected blood vessels. Symptoms include reduced vision, greater sensitivity to bright lights (photophobia), involuntary movement of the eyes (nystagmus) and strabismus.
https://albinismo.es/wp-content/uploads/CuadernoEducacion.pdf

- **Legg-Calvè-Perthes disease**

This is an uncommon disorder characterised by unilateral or bilateral avascular necrosis of the femoral head in children. The initial symptoms are limping, pain in hips, thighs or knees and reduced hip movement. Later, a discrepancy may be observed in the length of the leg together with muscular atrophy around the hip. The active phase of the disease may last several years and is characterised by the partial or total necrosis of the head of the femur and its progressive deformity. The final extent of the deformity may vary from an almost normal configuration of the joint to an extensive deformity.
https://asfape.org/publicaciones/manual-colegios/

➢ Noonan Syndrome* and other Rasopathies

Noonan Syndrome is a genetic condition with variable expressivity. A common feature of all rasopathies is the alteration in the regulation of an intracellular signalling pathway called the RAS-MAPK pathway, which seems to explain the widely different manifestations of these disorders such as deterioration in growth, heart problems, predisposition to develop tumours, among others. Other disorders that fall within the group of rasopathies include Neurofibromatosis Type 1, Leopard Syndrome, Costello Syndrome and Faciocutaneous Syndrome.
http://www.noonanasturias.com/educacion/

Neurofibromatosis «Psychopedagogical perspective»
http://www.acnefi.org/revista/n00760.htm

***** *Syndrome = a group of symptoms (pain or discomfort reported by the patient) and signs (clinical data observed by a doctor) that characterize a health condition.*

➢ Xeroderma pigmentosum

Xeroderma pigmentosum (XP) is a rare form of genodermatosis (dermatosis of genetic origin) characterized by extreme sensitivity in the skin and eyes to changes induced by ultraviolet rays (UV). From the first months of life affected children may be extremely sensitive to the Sun, resulting in serious burns and a high risk of developing melanomas.
https://xerodermapigmentosum.es/padres-y-cuidadores-2/protocolo-de-actuacion-en-el-centro-educativo/

➢ 22q11.2 Deletion Syndrome

22q11.2 deletion syndrome (DiGeorge syndrome) is caused by a chromosome anomaly. In general, affected people may suffer malformations of the heart, facial dimorphism, delayed development with or without intellectual disability, behavioural disorders, difficulty speaking due to poor functioning of the soft palate, problems with hypocalcaemia and on occasions, immunodeficiency.

Many children show moderate facial dimorphism (narrow palpebral fissures and bulbous nasal tip, small mouth and ears, malar hypoplasia) and orthopaedic anomalies (kyphosis, scoliosis, butterfly vertebra, hemivertebra, club foot, polydactyly).
https://www.22q.es/docs/files/7_gua-de-intervencin-educativa-22q11-aswillen.pdff

➢ Prader-Willi Syndrome (PWS)

Prader-Willi Syndrome is a genetic disease with a complex clinical expression that affects many different systems in the body. It is considered the most common cause of obesity of genetic origin. It is manifested at birth by a muscle weakness which leads to feeding problems in new-born babies. Later they develop hyperphagia but without ever feeling full. This is why they must ingest less calories than normal and must always follow a strict diet with regular physical activity. This syndrome is associated with a delay in development and learning difficulties, which mean they need support at school, as well as behavioural problems, a high pain threshold, sleep disorders and short stature.

Pupils with Prader-Willi Syndrome are hypersensitive and have trouble controlling their emotions. The frustrations related with this illness are in part responsible for their behaviour issues.
https://www.praderwillicat.org/wp-content/uploads/2019/11/GUIA-ESCOLAR-SPW-2019.pdf

https://www.praderwillicat.org/wp-content/uploads/2017/11/buenaspracticasaulas.pdf

- **Achondroplasia**

Achondroplasia is an alteration in bone formation which affects the growth plate of the long bones. It is of genetic origin and manifests itself in short stature, macrocephaly and other bone alterations. Sufferers develop normal intelligence and it may be associated with other health problems such as glucose intolerance, secondary hydrocephalus or spinal complications.
https://www.fundacionalpe.org/images/alpe/library/Carpeta-accesible-para-nios-con-acondroplasia.pdf
https://www.fundacionalpe.org/images/alpe/library/Orientaciones-escuela.-Primaria.pdf

- **Mastocytosis**

A disease caused by an abnormal increase in the number of mastocytes (cells that participate in allergic and inflammation processes and also affect the body's defences), in different areas of the body.

The organ most frequently affected by this disease is the skin, which presents red-violet cutaneous lesions. Other organs that may also be affected include the bone marrow, bones, liver, spleen and gastrointestinal tract.

Its clinical manifestations include itching, facial reddening, general malaise, blisters, abdominal pain, diarrhoea, difficulty concentrating, malabsorption, osteoporosis and anaphylaxis, among other problems. The mastocytes may be activated by strong contrasts in temperature (intense heat or cold) and intense friction or pressure on the skin or due to emotional factors: stress, anxiety.
http://www.elfarodetiza.com/articulos/patologia-mastocitaria-y-educacion-n251

- **Williams Syndrome**

This is a multisystemic genetic, neurodevelopmental disorder with a characteristic facial appearance, cardiac anomalies (the most common of which is supravalvular aortic stenosis), cognitive and developmental anomalies, and anomalies of the connective tissue (such as joint laxity).
http://www.sindromewilliams.org/wp-content/uploads/2017/09/Intervencio%CC%81n-educativa-SW.pdf

http://www.sindromewilliams.org/wp-content/uploads/2017/09/La-ansiedad-en-los-nin%CC%83os-con-SW.-Guia-educadores-primaria.pdf

NOTE: we are unable to include all the associations that offer support information for schools. We recommend contacting the corresponding associations.

MAP of RD Associations in Spain: https://creenfermedadesraras.imserso.es/creer_01/recuasoc/mapaso/index.htm

✓ 7.4 Rare Diseases and Behaviour

Some minority diseases are associated with behaviour problems: genetic syndromes such as Prader Willi Syndrome, Smith-Magenis Syndrome, some mucopolysaccharidoses such as Sanchez Syndrome, Sanfilippo Syndrome, Lesch-Nyhan Syndrome, Huntington Disease, Niemann-Pick Disease…. These problems must be treated as complications of the child's medical condition and not as a form of bad behaviour.

Hyperactivity, anxiety, fear and irritability are involuntary responses that interfere with learning, adaptation and the general classroom atmosphere. It is therefore important to establish cause-effect relations in order to be able to anticipate and control the appearance of these problems by making changes in the learning atmosphere, in methods, by using positive reinforcement, etc.

2nd Conference on Inclusive Education. RD and Behaviour (Creer)
https://www.youtube.com/results?search_query=II+jornadas++de+inclusi%C3%B3n+educativa+Conducta+y+enfermedades+raras

✓ 7.5 Other guides offering advice

In the classroom, we may also encounter other complex health conditions such as cancer, congenital cardiopathies which often correspond to or are associated with a minority disease.

Here are some guides that tackle various aspects and offer advice as to how to attend to pupils with these pathologies, their families and classmates.

Cancer in the classroom
https://www.afanion.org/bddocumentos/Gu%C3%ADa-de-Educaci%C3%B3n.pdf

Heart disease in the classroom
https://www.menudoscorazones.org/wp-content/uploads/2019/09/Tengo-un-ni%C3%B1o-o-una-ni%C3%B1a-con-cardiopat%C3%ADa-en-el-aula.pdf

8. FIRST AID FOR PUPILS WITH RD

The **Response Protocols in first aid and medical emergencies** drawn up by the Educational Administration for different health conditions and chronic diseases offer valid responses that can be extended to pupils with minority diseases.

CANARIAS
https://www.gobiernodecanarias.org/educacion/web/centros/protocolos/asistencia_sanitaria_alumnado/

CASTILLA Y LEÓN
Portal de Educación de la Junta de Castilla y León - Protocolos de actuación ante urgencias sanitarias en los centros educativos de Castilla y León. (jcyl.es)

ANDALUCIA
https://www.juntadeandalucia.es/educacion/portals/web/escolarizacion-segura/presentacion

https://blogsaverroes.juntadeandalucia.es/orientajaen/files/2017/08/Enfermedades_cr%C3%B3nicas.pdf

We also have **guides about first aid in rare diseases** drawn up by patients' associations and specialized health centres, such as:

- **West Syndrome and other epileptic encephalopathies**

West Syndrome is another name for infantile spasms and belongs to a group of conditions known as "catastrophic epileptic encephalopathies". Infantile spasms are a special type of epileptic attack which affects above all children of less than one year old.
It is classified as a catastrophic syndrome due to the fact that it has a death rate of around 5%. Patients often suffer side effects such as psychomotor delay, autism, hyperactivity and refractory epilepsy (the crises cannot be controlled and this prevents patients from leading a normal life)
http://www.sindromedewest.org/PDF/LIBROS/Gu%C3%ADa.pdf

- **Congenital Metabolic Errors**

Congenital Metabolic Errors (CME) form a very numerous class of uncommon diseases. They are caused by hereditary alterations in DNA called mutations, which generate anomalous proteins with an altered structure and therefore altered functions. This causes malfunctioning of cells and organs.
https://metabolicas.sjdhospitalbarcelona.org/consejo/nino-aciduria-metilmalonica-escuela-profesores-cuidadores

9. WHERE SHOULD I GO IF I HAVE A PUPIL WITH…?

✓ **Specialized Resources**

SCHOOL ADVICE SERVICES OFFERED BY PATIENTS' ORGANIZATIONS

Numerous associations, in collaboration with professionals who are experts at attending to people with different rare diseases, have developed advice programmes for schools and visit schools on request.

- National Association for Brittle Bone Disease (Ahuce) Osteogenisis Imperfecta
  http://www.ahuce.org/Servicios_que_presta_AHUCE/AsesoramientoEscolar.aspx

- Spanish Association for Familial Mediterranean Fever and autoinflammatory syndromes
  https://fmf.org.es/servicios/servicio-de-asesoramiento-en-el-entorno-educativo/

- Spanish Association for Cystic Fibrosis

  https://fqvalenciana.com/manuales/

  https://www.youtube.com/watch?v=BBUCLVdl-AA

SERVICES PROVIDED BY THE NATIONAL CENTRE OF REFERENCE FOR ATTENTION TO PEOPLE WITH RARE DISEASES AND THEIR FAMILIES (Creer)

Creer's Education Department is made up of a pedagogue, Yolanda Ahedo Infante, and two teachers, Montserrat Cabrejas del Campo and Ana Santamaría Herrera.

Schools or any of their **professionals** can seek **advice and guidance** as to the best ways to attend to pupils with rare diseases at school. They may also **apply to take part** in the "Programme for Raising Awareness and Dissemination in Schools" by any of the following means:

✉ **escuela@creenfermedadesraras.es**
947 253 950
C / Bernardino Obregón, 24
09001 Burgos

Creer Information and Advice Service

Service aimed at:

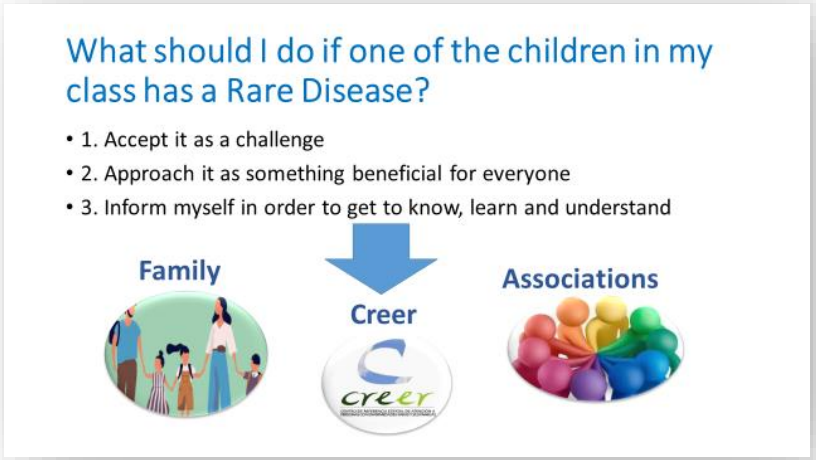
- ✓ People affected by a Rare Disease.
- ✓ Relatives of people affected by a Rare Disease.
- ✓ **Professionals who require information and/or advice.**
- ✓ The general public.

✉ **info@creenfermedadesraras.es**

SOURCES OF CLINICAL INFORMATION

The European Portal **Orphanet** offers clinical descriptions of numerous rare conditions and is the first source recommended for gathering information. It also contains data about patient support organizations, expert centres, essays and research projects, among other information of interest.
https://www.orpha.net/consor4.01/www/cgi-bin/Disease.php?lng=ES

The Metabolics Portal of the Hospital San Joan de Deu in Barcelona offers complete, easily accessible information about a range of different Congenital Metabolic Errors and resources to help children find out about and manage their illness, which may be useful for work in the classroom.
https://metabolicas.sjdhospitalbarcelona.org/enfermedades-metabolicas



Summary of the main ideas set out in sections 7, 8 and 9.

10. TESTIMONIES

➢ **THE MOST BEAUTIFUL THING IN THE WORLD**

Yesterday, my daughter came home from school in a terrible state. The pain in her back had once again tinged her face with sadness. Barely saying hello, she ran to her room to change her clothes, as she always does when she has a bad day. When at last she came into the living room, I asked her how she was, and she almost burst into tears. "Bad" she whispered. Her father and I exchanged impotent glances. The medicine was not having the desired effect and we had already tried so many different ones …

My daughter is normally a positive, happy child. When she was little, she was always laughing, in spite of her condition. She was born with a serious, very rare malformation, which marked our lives forever.

She was born with **Cloacal Exstrophy**. Her bladder had barely formed into two small pieces, which came out through a hole in her belly, through which the small intestine, shorter than normal, also peeped out. Her colon was "missing", she had no anus, no exit from her urethra, … and no exit to the outside as if she were a plastic doll. Her hips were so wide open that each leg was pointing away from the other (they thought she would never be able to walk). In her spinal column she had spina bifida occulta with sacral lipoma and tethered cord, her kidneys were joined together and out of place, one almost in the pelvis. 20 years later I still haven't recovered from the shock.

Like so many parents of children with rare diseases I had never heard about it before. I did not know that in the hospital where my daughter was born, they did not know how to treat it and nobody told me. After the first failed operations, we found another hospital nearby, where they didn't admit that

they didn't know about it either and where over the next several years she was subjected to more failed operations.

At last we reached the Hospital La Paz in Madrid, where they have specialists in this kind of malformation and nowadays they have a CSUR (a specialized centre of reference) for Exstrophy. We thought that at last we had found the help we needed… But **she had had so many operations that it was far from easy**. It took her several years to achieve artificial continence and she wore nappies until she was 12. They were unable to fix her intestine or conduct it to the anus.

Throughout this period, **she suffered terrible complications**: very serious bouts of peritonitis, intestinal obstructions, paralytic ileus, renal infections … and a long etc. which had a very negative impact on her quality of life and meant that **she spent a long time in and out of hospital.**

Even so, she was always happy. **She never fussed about the nappies or the ileostomy bag, or the limp** (which with the aid of physiotherapy was all that was left of the fear of not being able to walk). She made friends in hospital with those in the beds at her side; **at primary school she appeared in front of her classmates, and with the help of a letter and a sample of her catheters and her bags, she explained in her own words what the matter with her was**. That way she avoided curious questions (she was so small!).

She was always a good student in spite of missing so many classes (sometimes for months). She is very intelligent and clued up and is proud of being so. At the end of her secondary education, the Regional Government of Galicia awarded her a prize for "PERSONAL EFFORT AND OVERCOMING ADVERSITY" (highly deserved).

This period came to an end and there were fewer hospitalizations and trips to the doctor in Madrid, but her quality of life took a turn for the worse. She began to suffer

pains in her back. She had always had problems with hip pain due to their incorrect anatomical disposition, but only if she had to stand or walk for a long time.

This pain was new and more and more disabling.

We searched for all possible causes and there were various: the tethered cord, the position of her hip… the osteoporosis. Yes, she now had OSTEOPOROSIS too!!! It is caused by her short intestine which does not allow her to carry out a complete digestion, which means that her bones do not receive sufficient calcium. This in spite of the fact that in a bid to prevent this problem she has been taking calcium and vitamin D since she was a small child, but it was not enough.

Due to the risk of paralytic ileus, she cannot take certain painkillers, and the ones she can take are not sufficient. We tried various different treatments in the hospital's pain unit, including deep Botox injections but none of them worked.

While all this was going on, she entered adolescence. She was always very mature for her age and although she hardly had any social life outside the family, she never felt the need. She has a fantastic relationship with her older brother and they used to play video games together or go to the cinema, she didn't ask for more. But adolescence makes us feel lonelier, more different, weirder … and often sadder too. She is no exception.

She would express her pain and her frustration on social media and she would often find consolation there and some form of social life. And a few times a year she meets up with her friends from Asafex, the Exstrophy Association that we belong to, in which she can be herself and where they understand her better.

And why don't people understand her? Because my daughter looks great. She is very beautiful, it's not just a mother's biased opinion. **If you cross her in the street or if you sit next to her at the University, you won't notice**

anything. She has a recognized disability of 76%, but it is completely invisible to other people. And she has to put up with comments when she comes out of the disabled toilets, which she has to use to manage the catheters, and listen to whispered gossip if she sits down on the bus because she can't stand the pain while I remain standing. She also has to put up with people she knows who know about her illness, saying things like: "You're looking really well. You're cured, aren't you?"

No. No, she is not cured. She is just brave and tough and strong … Because she has never had a choice; she has had to learn to live with the pain, with the ileostomy bags, with the catheters for urinating which don't go in well, with continuous urinary infections, frequent dehydration, with a strange diet because of her many intolerances, with orthopaedic insoles that don't allow her to wear high heels or nice shoes, but without which she couldn't walk more than 10 paces, with the pills, the patches, the bladder washes, and the medical check-ups …

She is now in second year at University and going to class involves constant pain. But she tries hard not to miss class and to get good grades (I'm SO proud of her!!!)

That's why when she smiles (which is often), when she laughs at herself, when she jokes about her situation, without any bitterness … my daughter's smile is literally **THE MOST BEAUTIFUL THING IN THE WORLD.**

- **LETTER FOR JULIA**

Dear Julia: Of the dozen and a half letters I have written for you, this is perhaps the one that I have found most difficult to write. There are just a few days to go now before you begin a new stage in your life and ours…., a stage which as in many others, we hope to be able to accompany you. We have left behind this summer, the days spent camping, at the pool, days

full of laughter and good fun, days at the hospital, days for making friends, for enjoying …. and in no time at all ….
YOU ARE STARTING SCHOOL!!!!! I can't say it any louder, but I am going to say it again: YOU ARE STARTING SCHOOL!!!!! **Wow!!!.**

We want you to know that we are a bit nervous, we are feeling a bit odd and slightly disconcerted, but I want you to know too that we are very happy and very excited to watch you grow and to see that we are gradually finding some kind of normality and a little bit of peace in such a short life.

We have to tell you, although some things you know already, that **getting here has not been easy**, that we've been through good times and bad. You were born on 26th June at 9:40 in the morning. You weighed just 2 kilos and you were 43 cm long. You were already determined to leave your mark on the world, although we hardly knew how to hold you. You were so small … We went home from the hospital with you on 2nd July, with a whole jumbled mixed bag of emotions. At that time, nobody knew how much we would have to fight for you and the strength that you have always transmitted to us is something that I will admire about you for the rest of my life. You never stopped smiling.

During the first few months we began to suspect that something was not quite right and on 8th October (I remember because we were taking you home from hospital), we received a call in which they gave your illness a name: **Russell-Silver Syndrome.** By that time, we had already begun all kinds of treatments that would improve your development as much as possible, and by that time, **in less than eighteen months, you had already been admitted 5 times** and your mother had been fighting it out with all the health professionals. Your father calmed my anger, shared my fears and transmitted a level-headed serenity that was also worthy of admiration.

Three years have now gone by, three exhausting years, more than exhausting, however three years of our lives in which there has always been something wonderful, without strange unusual fears, and that was you. You are a constant bundle of joy for every little step you take and every little thing you achieve. I want to tell you that we have reached this point, always together, and that fear, insecurity or sadness are just some of the emotions that we normally feel when faced with any change. But, at this stage of the game, our fears are gradually being left behind and hope is taking their place. **With this new thought, we come to the end of 2015, which is when we decided that you should go to school**. **Even though you still seem so small and vulnerable,** and at the same time so big and such a fighter. Your Mum, not so much your Dad who was always more convinced than I was, experienced her first doubts and uncertainties: **I debated whether to send you to school or not.** You've never been to infant school because **your life has always been accompanied by infections and periods of illness,** which made it difficult for you to attend any form of preschool. You weigh just 9 kg and as of today any infection would mean being admitted to hospital again, but I thought about you and I saw that look on your face and I saw that beyond all this, the time had come. I realize that you need to establish other bonds outside the adult world. We have been resolving all these doubts with the help of some people who know you very well: Berta (Social worker at GERNA), your paediatrician (a vital piece of the medical puzzle), Elena Ganuza ("Elena the lady for playing" as you call her) and the Primary Care team. Without forgetting our friends and family who have been encouraging us and providing a little peace. We all agree that you have to go to school (it's strange, but now that I am writing to you, I like to think that you agree too), but we have to weigh everything up carefully, to see and to plan how to do it so that **the change disrupts your life as little as possible.**

You are a girl who emanates something special but we cannot forget that you are a girl of three years old in a body of two, who has certain motor difficulties and a slight delay in terms of language. You are also a girl who never feels hungry and as a result **you eat through a gastrostomy tube or gastric button and it's necessary to control your sugar levels.**

As you will see when you read these letters, this was a very important decision that we had to take. My darling daughter, for you we have chosen the «El Lago de Mendillorri» School. We thought about this school because your cousins studied there and they were always very happy, because it's very near home, and **because it's near the health centre. Wow, what an important reason for choosing a school...** Apart from the resources and needs that I will be telling you about later, given your medical profile and the fact that we can't have a nurse at school, having the health centre nearby with your paediatrician, Mari José, your nurse, Iris, and the girls from the office who love you to bits, well it's very important for us because it makes us feel more calm and relaxed. We can't always arrive on time to rescue you and we don't always know how to. So far so good … The second part was to ask for or find out what type of resources you will need to make the most of your time at school. **We think it's just as important to give you the support you really need as not to give you more than you really need.**

At the beginning, **the Education Department has decided that you be accompanied by a carer, a speech therapist and all the necessary adaptations to the furniture that allow you to go about your day in a safe environment**. Now that all the puzzle is complete, I think you should know that you have been enrolled and that during the summer, in which we've had such a great time with you, we've been telling you and letting you know a little about what's going to happen and we've talked about school, that you're going to

be playing with other children, and so far it seems like you like the idea, you smile when we talk about it. Mum and Dad have also been organizing themselves. It's good for us to prepare ourselves too and sort out all our little things and our questions.

When a child with an uncommon disease arrives at school, as in your case, daughter, **she arrives with a report**, which describes more or less accurately her current stage of development. **The report doesn't have much information about your disease or syndrome and what it involves.** This is why **we as your parents feel the need to explain as effectively as possible the details of your illness** although we are also worried about: How do we ensure that while you are different, you aren't …? How do we explain to your classmates that you are just as keen to do things as they are and that often you don't do something not because you don't want to but because you can't? How do we explain to the whole educational community that you are children who often lack the ability to communicate, that you are or often feel insecure, and that you express a great deal of frustration because you are incapable of resolving conflicts effectively. It is so important for people to be observant and a look is so important for you …

I have prepared your backpack already. It's so hard for me to take this step … But I know you will be happy. We hope not to have to fight too much against the system. I'm afraid of the wear and tear this new situation might entail. But we have the support we need and we hope to know how to do things well. So all we have to do now is wait for your big day to arrive.

Remember Julia… something about you: **IT'S THE DISEASE THAT'S STRANGE, NOT YOU.** We love you so much. Mum and Dad.

- **CONGENITAL NEVUS**

Hi, My name's Inma, and I'm the mother of a six-year-old girl with a giant congenital Nevus.

What's that? It's a dark brown mole or plate, a malformation of the skin, that covers part of the body, and requires dermatological care, surgery and physiotherapy.

We first encountered this problem when she was born in a natural birth. That moment is etched on my mind, the feeling of impotence at not being able to be with her, as they took her away for observation.

At that time, **the doctors did not explain the situation** to me or my husband. The next day, the paediatrician told us that it looked to him like a nevus and that he hadn't seen a case like that in the previous 25 years, or with those characteristics. Every time I changed my daughter's nappy, **I felt guilty.** What have I done wrong? Little by little I came to accept it and gathered up the strength to help my daughter get through it. I fought and continue to fight to find out more about this disease, as the doctors did not have exact answers to the questions I was asking. Some were sincere when they told you that they would study the case, others just talked complete drivel, which at times like this makes you hit rock bottom. I also think that some of them should take a course in humanity.

Having a rare disease means going from specialist to specialist without receiving answers **and getting more and more frustrated**. Many thanks to the paediatrician and the dermatologist who were on duty that night, who have provided enormous moral and emotional support as they have got actively involved in the illness. My questions began to receive answers.

Soon afterwards, I came across the Asonevus Association, who informed me about most aspects of the illness and I also met some friends who understand the pain and

anxiety you are experiencing and give you the strength to continue with this war.

As they start telling you about all the risks this disease entails, your head starts thinking it's impossible and you begin to let yourself sink into a hole. But when that happens, I look at my daughter and she looks so pretty and so special and that gives me the strength to enjoy her as she is, that's the way she is, like my other daughter. Each one of us is different, some are tall, some are short, etc.

The toughest thing is when she asks me or tells me about things that have happened to her: "Why have I got this and my sister hasn't?", and "the other kids say I'm dirty". At moments like that, you get a knot in your stomach and you try to make her understand that what she has is special, but it doesn't make her different from other kids.

She is a very happy girl, who always has a smile on her face. She is full of joy!

- These testimonies were published in the Digital Magazine published periodically by Creer.

Picture of a collage with hands, hearts and other coloured shapes on a black background, with a text that reads: "In my school, there is space for everyone. Together we grow better". Made by the children from the CEIP Fuentecillas primary school in Burgos, on the occasion of World Rare Disease Day (29th February).

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