ALBINISM: a genetic condition

"Do not stay in the shadow"

French Albinism Association
Albinism...and now what?
Your child has just been diagnosed with albinism. And now what?

Now, there is life.
The diagnosis leaves you with many unanswered questions:
• What does my child see?
• What are the practical implications on his future life and on the one of our family?
• How will he learn to read and write having a visual impairment?
• What job will he be able to do?
• Will he pass this condition on to his children?
• If I have other children, will they be affected by albinism?
• What can medical research do?
We have asked ourselves all these questions and many others. We live with these questions every day.

Contact

Do you need advice? Do you have questions? Do you want to talk about albinism to someone? If so, do not hesitate to contact us!

If you live in France or in a neighboring French speaking country, feel free to contact directly the French association Genespoir.
• by telephone: +33 2 99 30 96 79
• by e-mail at info@genespoir.org
• by mail at Genespoir, 3 rue de la paix, 35 000 RENNES - France

If you are looking for more information about the association, you can visit the website www.genespoir.org
If you are not a French speaker and/or you want to contact Albinism Europe, feel free to reach out to:
Antoine Gliksohn (in charge of the international affairs)
e-mail: international@genespoir.org
www.albinism.eu
The doctors of the multidisciplinary consultation on albinism of the Bordeaux reference center
Pr Alain Taïeb, dermatologist
Pr Benoît Arveiler, geneticist
Dr Clément Paya, ophthalmologist
as well as
Dr Catherine Duncombe Poulet, ophthalmologist specialist on strabismus
Dr Alain Spielmann, ophthalmologist specialist on strabismus
have contributed to draft this brochure in French.

The text has been translated into English with the help of Nicola Birkbeck, Rosaleen Dempsey (the Albinism Fellowship UK & Ireland) and the US organization NOAH.

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The patients’ organization Genespoir
Genespoir was created in 1995 in Rennes, France, by Fabienne Jouan, a mother of three children, two of them having oculocutaneous albinism. Today, it is the main French patients’ organization specific to albinism.
The association has over 350 members: adults and children with albinism, parents and relatives of children with albinism, friends and acquaintances. Several of these members live outside France, in the francophone regions of Belgium and Switzerland, as well as in Luxemburg.
Genespoir informs the families as well as the general public and the medical professionals about albinism and its consequences: diagnosis, skin protection, visual aids, social, scholar and professional inclusion... The association provides families psychological assistance and informs people with albinism on their rights. Genespoir actively promotes scientific research on albinism. It collects donations from private citizens and organizes fundraising events in order to provide financial support to research programs.
Genespoir acts so the French public health care system takes charge of treatments. Likewise, it represents and defends the interests of every person with albinism and devotes significant attention to break the isolation barrier of people with albinism thanks to the organization of exchanges between the members of the association.

Albinism Europe
Genespoir is a member of the European network of patients’ organizations called “Albinism Europe”.
At this time, ten associations are part of this initiative :
• Denmark - Dansk Forening For Albinisme - www.albinisme.dk
• Finland - Suomen Albinismiyhdistys ry - http://albinismiyhdistys.omasivu.fi
• France - Genespoir - www.genespoir.org
• Germany - NOAH Albinismus Selbsthilfegruppe e.V - www.albinismus.de
• Italy - Albinit - www.albinit.org
• Netherlands - Oogvereniging Albinisme - www.oogvereniging.nl/ledengroep/patientengroep-albinisme/
• Norway - Norsk Forening For Albinisme - www.albinisme.no
• Spain - Alba - www.albinismo.es
• Turkey - Albinizm Derneği - www.albinizm.org.tr
• UK & Ireland - The Albinism Fellowship - www.albinism.org.uk

Up to now, the organization has agreed on four top priorities to act at the European level
• Exchanging experiences and best practices between the European associations as well as sharing information
• Promoting best practices in the medical and educational fields as well as acting for the development of international guidelines on albinism
• Stimulating the research and connecting as far as possible the scientists working on albinism
• Joint awareness-raising campaigns and public relations activities
One of the most successful achievements of the network is the organization of the **European Days of Albinism**, a conference on albinism dedicated to researchers and medical practitioners as well as to representatives of patients’ organizations. After four conferences successively organized in Paris (France) in October 2012, in Valencia (Spain) in April 2014, in Milan (Italy) in April 2016 and in Hurdal (Norway) in March 2018, we are getting ready for a fifth conference planned for the spring of 2020 in France.

Partly thanks to the financial support of the ERASMUS+ program, Albinism Europe has also been organizing meetings dedicated to young people with albinism coming from all over Europe.

Since January 2017 the organization has established a Scientific Committee made of the following international experts on albinism:

- **Benoît Arveiler** (Bordeaux, France)
- **Lluís Montoliu** (Madrid, Spain)
- **Brian Brooks** (Bethesda, USA)
- **Maria van Genderen** (Zeist, Netherlands)
- **Irene Gottlob** (Leicester, UK)
- **Karen Grønskov** (Copenhagen, Denmark)
- **Michael Hoffmann** (Magdeburg, Germany)
- **Barbara Käsmann-Kellner** (Homburg, Germany)
- **Alessandra Del Longo** (Milan, Italy)
- **Michael Marks** (Philadelphia, USA)
- **Fanny Morice-Picard** (Bordeaux, France)
- **Vittoria Schiaffino** (Milan, Italy)
- **Maria José Trujillo** (Madrid, Spain)
Introduction
Albinism is a genetic condition which people inherit from their parents and can pass on to their children.

The main characteristics of albinism are vision defects: hypopigmentation of the retina, iris transillumination, underdevelopment or absence of the fovea and abnormal crossing of the optic nerve fibers. The consequences of these defects are reduced visual acuity, nystagmus, light sensitivity and possible squint.

If the symptoms are limited to these visual defects, we are referring to ocular albinism. However, if they are also accompanied by a general hypopigmentation of the skin and the hair (with varying degrees), we are referring to oculocutaneous albinism. More rarely, albinism is associated with other symptoms such as abnormalities in the blood cells, lungs and intestines.

All in all, albinism affects around 1 in 17,000 persons in Europe.

In any case, it is caused by an abnormality in the production of a pigment: melanin.

Different forms of albinism
There are many types of albinism, and these types can be classified either according to the phenotype (i.e. the visible differences in individuals) or according to the genetic origin (i.e. the gene involved).

The medical community is attempting to determine and understand the links between genotype and phenotype, but these links remain somewhat poorly understood. Indeed, it is very difficult to understand the correlations between genotype and phenotype. For instance, we often note that different mutations on the same gene can lead to extremely varied phenotypes. Likewise, mutations on different genes can sometimes generate similar phenotypes.

The different phenotypes
Oculocutaneous albinism
Oculocutaneous albinism refers to a form of albinism in which visual defects are accompanied by a reduction or almost complete absence of pigmentation (melanin) in the skin and hair. This lack of pigmentation gives affected people a particular physical appearance, which is often well known by the general public.
Ocular albinism

Ocular albinism refers to a form of albinism in which visual defects are the only consequence of the patient’s genetic abnormality. The skin and the hair seem to have a normal pigmentation, although in some cases the patient’s pigmentation may appear slightly lighter than the rest of the family. There is a particular form of ocular albinism which is notable for the fact that the iris and the retina of the patient display a normal pigmentation, while the other ocular characteristics of albinism are present. This form of albinism is called FHONDA Syndrome: Foveal Hypoplasia Optic Nerve decussation Defects and Anterior segment dysgenesis.

The forms of Syndromic albinism

People with these rare forms of albinism present the typical ocular characteristics of albinism. Their phenotype can be either only ocular or also cutaneous. Moreover, these people are concerned by other additional specific medical conditions affecting other organs such as the intestines, lungs, blood platelets or the immune system.

There are two types of syndromic albinism: Hermansky-Pudlak syndrome (HPS) and Chediak-Higashi syndrome (CHS).

Hermansky-Pudlak syndrome

In the case of this syndrome, albinism is systematically accompanied by bleeding disorders due to a dysfunction of the blood platelets. These disorders express themselves by a slower blood clotting. That implies prolonged bleedings in case of injuries as well as a bleeding tendency in general (nose or gums bleeding). Besides, bruises are very common even after mild bumps. Furthermore, serious complications such as hemorrhage may occur during surgery or delivery.

Medications having anticoagulant properties, such as aspirin, and non-steroidal anti-inflammatory should be avoided at all costs.

In some cases, a chronic intestinal inflammation (colitis) or a pulmonary fibrosis (lung) may occur.

In very rare cases, a renal failure, a cardiomyopathy can be noticed. Likewise, a decrease of the white blood cells (neutropenia) can appear and increase the risk of infection.

Daniel, 64 y/o, HPS5, 20/200 (or 0.1)
Chediak Higashi Syndrome

The cases of Chediak Higashi Syndrome are extremely rare (around one case out of one million births). The persons affected have, as in the case of HPS, a form of albinism and a dysfunction of the blood platelets. But in addition to these symptoms, neurological disorders and serious immune deficiencies are to be noticed. Therefore, patients suffer frequent infections. Currently, the only known treatment is a bone-marrow transplant which should take place during the very early years.

“I don’t think I am disabled because of my albinism. It is in other circumstances that I experienced limitations: – when I jumped into the venture of the restaurant for example, and people didn’t take me seriously because I was young.”

Guillaume, head cook

Photo: Anna Delachaueme

Different genetic origins

People’s physical characteristics are defined by the genes they have inherited from their parents. These genes are carried by chromosomes. Each human cell contains 22 pairs of homologous chromosomes (autosomes) and one pair of sex chromosomes which can be of two types: X or Y. Females have a pair of X homologous chromosomes, while males have one X chromosome and one Y. Except for the genes carried by the non-homologous X and Y chromosomes, each of our cells contains two copies (allele) of each gene. One comes from the father and the other one from the mother. There are dominant alleles and recessive alleles.

The variety of genes

In 2017, we know of nineteen genes which could be responsible for different types of albinism (see Table below). Most of them have been discovered in recent years and there are good reasons to think that other genes remain to be discovered.

The first gene discovered was the TYR gene. In France, this gene is responsible for the greatest number of albinism cases. When the TYR gene is mutated, we are referring to type 1 oculocutaneous albinism (OCA1) since most of the time, the patients present an oculocutaneous phenotype. Nevertheless, a dysfunction of the TYR gene can also lead to a phenotype which apparently looks only ocular. And that doesn’t only apply to the TYR genes, but more generally, to the OCA2, TYRP1, SLC45A2, SLC24A5 and C10orf11 genes, which are associated to the types of albinism OCA1 to 7. In other words, for all of these genes, the degree of hypopigmentation can vary, from a total absence of melanin in the skin and the hair to a relatively normal pigmentation.
When they are defective, 11 out of the 19 known genes linked to albinism lead to syndromic forms. Today, we can identify 10 types of HPS and one single type of CHS. Depending on the mutations, the person affected by a syndromic form may receive a diagnosis of either oculocutaneous albinism, or ocular albinism. The GRP143 gene is the only one out of the 19 albinism genes to be situated on a sex chromosome (X chromosome). It exclusively leads to ocular albinism. At this respect, some doctors commonly call this form X-linked ocular albinism. Except in exceptional cases, this form of albinism only affects boys.

Finally, the SLC38A8 gene stands apart from the other albinism genes as it doesn’t cause any defect of pigmentation at both ocular and cutaneous level when it is mutated. In other words, all the people having this dysfunction present the characteristics of ocular albinism without nevertheless any hypopigmentation. This particular form is called FHONDA syndrome.

When a patient presents a form of albinism, clinical examination (observation of the phenotype) is not enough to identify the gene involved. Only a molecular diagnosis (genetic) can conclusively tell us the form of albinism in question. The molecular diagnosis especially allows us to distinguish the classic forms of albinism from the syndromic forms, which require specific medical attention. It is also extremely useful in the case of mild forms, where clinical results leave room for doubt as to whether the patient is affected by albinism.

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<tr>
<th>Gene</th>
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<td>OCA4</td>
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<tr>
<td>n.d.</td>
<td>OCA5</td>
<td>Oculocutaneous Albinism Type 5</td>
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<td>OCA6</td>
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<td>OCA7</td>
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<td>OA1</td>
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<td>SLC38A8</td>
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<tr>
<td>AP3D1</td>
<td>HPS10</td>
<td>Hermansky–Pudlak Syndrome Type 10</td>
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List of all known genes responsible for the different types of albinism in 2017.
The known modes of transmission
There are two different modes of transmission: autosomal recessive albinism and X-linked albinism. All known genes are of the autosomal recessive type, except GRP143, which is linked to the X chromosome.

Autosomal recessive transmission
In this case, the person affected carries two defective alleles on the same gene. Both parents, who are not affected by albinism themselves, carry a defective (recessive) allele and a dominant functional allele. So, statistically, at each birth, the probability for the child of inheriting the changed alleles from both parents and thus being affected, is one in four. This is the most frequent mode of transmission. The probability that two unaffected parents are carriers of the same defective allele of a given gene is high if they have common ancestors (consanguinity). This increases the risk of them having a child with a genetic condition such as albinism.

X-linked transmission
In this case, a non-affected woman carrying the unique changed allele of the albinism gene has a 1 in 2 chance of transmitting this copy to each of her children. Every male carrier of this changed gene will be affected by ocular albinism and will pass this gene on to all his daughters. His daughters will be affected only if their mother carries this same gene, which is extremely rare. This situation can especially happen if both parents are relatives.
Albinism and Vision

In temperate regions, the main effect of albinism is the alteration of the visual function.

Normal Visual Function

Light enters the eye through the pupil, which is the opening at the center of the iris. The iris controls the amount of light entering the eye by either contracting or dilating. The visual function begins at the back of the eye on the retina, upon which the image is formed. The retina is made of several layers of cells: the layer of nerve fibers and the layer of retinal fibers, both transparent; the layer of photoreceptors, which receive the light; the layer of retinal pigment epithelium, where melanin is normally stored (fig. 1); the choroid, which contains blood vessels; the sclera, which is the white covering of the eyeball.

All the visual information the retina receives is transmitted to the visual areas of the brain by the optic nerve. The macula is a zone situated in the center of the retina, in the axis of the pupil. At its center, there is an area measuring approximately 2mm in diameter, which is slightly concave: the fovea. Here, the density of the light receptive cells reaches its maximum. This is in this area that the retina shows the best visual perception (100/100). The surrounding areas of the retina don’t allow to see well the details, their acuity is very much lower, but it is useful in the perception of space and movement.
**Visual Function of people with albinism**

The eye of a person with albinism has an iris more or less transparent; the foveal depression, the hollow at the center of the macula, is more or less absent, this is called macular hypoplasia\(^{(15)}\).

The absence of pigmentation in the iris and the retinal pigment epithelium is the main cause of photophobia. The absence of fovea is responsible for the weak visual acuity. If the fovea of a person affected by albinism is compared to the light sensor on a camera, it could be said that this fovea presents far less pixels than on a normal fovea.

**In a person affected by albinism, the optic nerves linking the eyes and the brain do not intersect in the usual manner.**

The eye and optic nerve’s anomalies that a person with albinism has prevent a normal maturation of the visual function and are believed to be responsible for the presence of a congenital nystagmus, which appears during the early months of life.

The weak visual capacity of the retina limits visual development and can cause strabismus, either convergent or divergent.

In addition to these disorders, extreme cases of astigmatism\(^{(16)}\) (usually due to anomalies in the sphericity of the cornea), hyperopia\(^{(17)}\) (far sightedness) or myopia\(^{(18)}\) (short sightedness) are frequent.

The visual impairment can be profound (acuity less than 10/100) but is more usually mean (acuity greater than or equal to 20/100). Color vision is usually normal.
The Physical Appearance of albinism

A person with oculocutaneous albinism usually has a particular physical appearance characterized by the absence or reduction of pigmentation of the skin, hair and body hair.

This phenomenon is particularly striking in populations where pigmentation is naturally high (persons from Africa or Asia) and is in these people, the most evident criteria for the diagnosis of albinism.

The degree of hypopigmentation can widely vary, depending on the altered gene but also on the type of changes.

The physical appearance of most of the people with albinism exposes them to the attention, stares and judgment of the public, while the crucial visual impairment is often completely ignored.

Beyond the visual problems they cause, nystagmus and strabismus can be troubling from an aesthetic point of view and can have a negative impact on social interactions. People may feel ill at ease when they are trying to make eye contact with a person with albinism because of the strabismus and the nystagmus. This can lead to problems in building rapport as people with albinism are often aware of their discomfort.
Medical Care

Diagnosis

Clinical diagnosis
The diagnosis of albinism can be made at birth if the clinical symptoms (hypopigmentation of the skin and hair) are obvious. However, in many cases, the hypopigmentation of the eyes, skin, hair and body hair is not sufficiently characteristic to immediately establish a diagnosis of albinism. It is during the first months of life, when nystagmus appears, that an ophthalmologic consultation is suggested.

The simplest examinations which should be initially performed consist in an examination with a slit lamp and an eye fundus (by ophthalmoscopy or fundus photography). To confirm the diagnosis and especially the macular hypoplasia, it may be necessary to perform an Optical Coherence Tomography (OCT). Complementary examinations, such as an Electretinogram (ERG), or Visual Evoked Potentials (VEP) can be prescribed. A VEP examination is not always easy to perform on a very young child.

As a precaution, quite a few doctors prescribe an MRI scan (magnetic resonance imaging). Yet, in the case of albinism, it does not provide any extra information. This test is pointless if the characteristics of albinism have been already identified by other ways. It should be noted that in order to carry out an MRI scan on a baby, a general anesthesia is necessary.

Molecular diagnosis
A molecular (or genetic) diagnosis is important. It allows to confirm, clarify and complete the clinical diagnosis. Besides, it allows a conclusive distinction between the classic and the syndromic forms of albinism, which is necessary to prescribe the most appropriate treatment. It is also particularly useful in cases of mild forms of albinism where the results of clinical examinations leave room for doubt.

Only a molecular diagnosis allows the families to receive a genetic counselling and the couples who want children to obtain precise information.

The current research allows us to anticipate possible treatments. They will be based (for each patient) on the knowledge of the gene and mutations causing albinism. Only the molecular diagnosis will produce guidance for patients and, at the right time, allow to suggest them to enter clinical trial protocols.

Finally, let’s remember that a molecular diagnosis is a right and that in France, it is totally covered by social welfare.
The diagnosis of all of the known genes of albinism can be done by the Laboratory of molecular genetics of Bordeaux University Hospital, led by the Professor Benoît Arveiler.

Address for a genetic diagnosis of albinism in France
Prof. Benoît ARVEILER
Laboratoire de Génétique Moléculaire
Plateau Technique de Biologie Moléculaire
Hôpital Pellegrin - 1er étage
1, Place Amélie Raba Léon - 33076 Bordeaux - FRANCE
Phone: +33 5 57 82 01 96

“Let me address the young parents: do not worry; they will surprise you, your little ones... but do everything you can to help them”
Jean-Marie, retired pharmacist

Albinism Day Hospital Schemes in Europe
Due to the wide variety that exists in terms of genotype and phenotype among people with albinism, it is recommended to carry out a clinical multidisciplinary evaluation of the condition for every patient. It allows in particular to establish an accurate clinical delineation of the phenotype and helps to define correlations between the genotype and the phenotype, which is essential to improve the general knowledge on albinism.
In this context, an Albinism Day Hospital Program has been implemented in several European hospitals over the past decades. This multidisciplinary consultation combines dermatological, genetic and ophthalmological assessments all scheduled over one day. Today Albinism Day Hospital Programs of Schemes are available in the following European hospitals:

• France: CHU de Bordeaux
  Secrétariat des Consultations de Dermatologie
  Phone: +33 5 56 79 56 22
  Email: christine.dambon@chu-bordeaux.fr

• France: Hôpital Necker-Enfants Malades à Paris
  Secrétariat de la consultation
  Phone: +33 1 44 49 46 62 or +33 1 44 49 43 37

• Italy: Milan Niguarda Ca’Granda Hospital
  A.O. Ospedale Niguarda Ca’ Granda – Milano Oculistica pediatrica:
  Phone: +39 02 6444 3297
Operations for nystagmus and strabismus

Nystagmus reduces the level of visual acuity and makes the process of seeing uncomfortable. Often, in order to improve their sight, the affected persons unconsciously adopt an abnormal and often uncomfortable posture in which the head is held at an angle. In this position called the null point, the nystagmus is reduced and sometimes almost stopped. It is important to note that the image perceived by a person with a congenital nystagmus (21) does not move, except in rare cases called oscillopsia (22). Nystagmus which may be related to strabismus may also be an obstacle to social interaction.

A nystagmus surgery is possible at the same time as a surgery to treat a potential associated strabismus. This surgery helps to lessen fatigue and to reduce visual discomfort, even if the gain in visual acuity is uncertain. In any case, such gain will be always limited by the deficiency of the retina. Furthermore, social relationships will be improved by ridding the other person of the previous embarrassment. At last, the related neck pain will also disappear.

In infancy, the earlier the surgery is performed, the greater the chance of further visual development is, which gives hope for a better reduction of the amblyopia in adulthood.

It is possible to operate from the age of two or three. It is also possible to perform this surgery on adults. There is no absolute rule as each case is unique.

Living with albinism

Protecting the skin

The natural body pigmentation protects people against the harmful rays of sunlight. For people with albinism, the lack of pigmentation makes them highly sensitive to the sun.

The pigment, the melanin, is produced in the melanosomes within a specialized group of cells known as melanocytes. Melanocytes are present in the skin, the hair, the pigmented layer of the retina, the iris and even in the inner ear.

In absence of this natural cutaneous protection against the sun, the main complications of oculocutaneous albinism are actinic keratosis (23), carcinoma (24) (either basal cell carcinoma (25) or epidermoid carcinoma; also called spinocellular carcinoma (26)) on the areas of the skin exposed to the sun. Cases of melanoma have been observed in patients with albinism, but their frequency does not seem higher than in the general population. Protecting the skin from sunlight from an early age can limit the development of skin lesions. The primary protection is clothing. Tops with long sleeves and long trousers made with a sufficiently opaque weave which limit the flow of UV rays are highly recommended (there are brands producing specialized anti-UV summer clothes, certified SPF 50+), as well as hats with wide brims. Moreover, the skin surfaces that remain exposed to the sun (face, hands, etc.) must be protected by anti-UVA and anti-UVB sun creams with a high SPF factor, following the recommended quantity and application frequency. Teenagers should be taught about these precautions.
and an annual medical check-up for precancerous and cancerous skin lesions is recommended. If these protection principles are respected, the risk of skin cancer is not higher than in the general population.

**Visual aids**
Alongside the other visual impairments, defects in the refraction of the eye (long sightedness, short sightedness, astigmatism) are to be noticed. These ones should be corrected as early as possible with glasses or hard contact lenses which filter the UV light.

Hard contact lenses allow an optimal optical correction as well as an excellent correction of astigmatism and often generate a reduction of the nystagmus, what also improves the central vision. Vision correction by continually wearing hard contact lenses\(^{(27)}\) is possible from a very early age (even before 6 months). It improves the development of the visual system, which is still immature in the first years of life. The optical correction must be the greatest possible to allow the best development of the baby’s or child’s vision. Photophobia can be eased and the retina can be protected wearing hats, sunglasses, or tinted contact lenses. Polarized glasses are recommended. Doing so, functional signs of visual defects can be significantly reduced and vision can be improved.

However, the ophthalmological problems due to the nystagmus, the strabismus, the foveal hypoplasia and the abnormal crossing of the optic nerve fibers cannot be corrected by glasses or contact lenses.

Visual aids (magnifying glass, digital magnifier and CCTV magnifier) can be used to improve near-vision in particular at school or at work.

Teenagers often don’t want to look different from their peers and refuse to use eye-catching aids. They can be offered discrete and efficient visual aids such as a pocket magnifying glass with a light for near vision and a monocular for distance vision.

**Inclusion at school**
The education of the relatives is essential. First, it involves informing those in charge of the educational follow-up of the child regarding his visual impairment. It will allow him to take simple measures in order to improve his everyday comfort and his inclusion. The child must sit as close as possible to the blackboard. Printed documents must be adapted (contrast, dimensions). In France, the institute for visually impaired children can appoint a specialized educator who will help the teacher in the classroom. The visual impairment must be mentioned in the child’s academic record. The child can also obtain an adjustment of his time schedule, enlarged documents as well as an adaptation of the time allowed to take national exams either in secondary education or in higher education. The use of an inclined desk or table can facilitate the child’s daily school life.
In France, according to the departments, different organizations can provide assistance to parents and teachers:

- **S3AIS**: a service to help and foster autonomy and social inclusion
- **S3AS**: a service to help having autonomy and for schooling
- **SAFEP**: a service of family support and for early education
- **CRDV**: the rehab center for people visually impaired of Clermont-Ferrand
- **IRECOV**: the rehab and educational institute for communication, hearing and sight of Tours
- **CAMFP**: a center for early medicosocial action

“When we practice judo, we forget the visual impairment”
Solène, 2010, 2012 and 2013 France handisport vice-champion in judo
Glossary

(1) **Hypopigmentation**: weak or absent coloration due to a lack of melanin which normally pigments the skin, the eyes and the hair.

(2) **Transillumination**: examination which consists of applying a light source on the side of the eye, observing then the light passing through the iris.

(3) **Fovea**: central area of the macula, where the vision is the sharpest.

(4) **Amblyopia**: significant decrease of the visual acuity without functional impairment.

(5) **Nystagmus**: succession of involuntary and saccadic eye movements, most of the time horizontal.

(6) **Photophobia**: visual discomfort due to an excess of light entering the eye.

(7) **Strabismus**: convergence defect of the visual axes preventing the binocular vision (simultaneous sight of both eyes). It can converge or diverge. In a child, it prevents the normal development of the stereoscopic vision (vision in relief or three-dimensional). Its origin should be rapidly investigated to rectify it.

(8) **Homologous chromosomes**: chromosomes which have the same form, the same size and the same genetic content.

(9) **Autosomal chromosomes**: chromosomes not involved in the sex determination.

(10) **Allele**: an allele is a version of a gene. There are several possible alleles of a single gene. Some alleles are functional, others defective.

(11) **Dominant allele**: a dominant allele only needs to be present in a single copy to express itself. It’s the contrary of the recessive allele.

(12) **Recessive allele**: a recessive allele should be transmitted by both parents to express itself in the case of autosomal genes. It’s the contrary of a dominant allele.

(13) **Autosomal allele**: it is transmitted by a chromosome from 1 to 22, except the X and Y sex chromosomes.

(14) **Macula**: yellow spot in the center of the retina. This is the area where visual acuity reaches its maximum.

(15) **Macular hypoplasia**: congenital insufficiency of the macula’s development.

(16) **Astigmatism**: vision defect due most of the time to anomalies respecting to the cornea curve.

(17) **Hypermetropia**: vision defect which generates difficulties to clearly see near objects.

(18) **Myopia**: vision defect which generates difficulties to clearly see far objects.

(19) **Genotype**: all the genetic characteristics of a person. We can consider the genotype as the “genetic identity card” of an organism.

(20) **Phenotype**: the phenotype refers to all of the visible characteristics of an organism: colour of hair, eyes, ears and nose shape, height, blood type… It is considered that the phenotype partly is an expression of the genotype.

(21) **Congenital nystagmus**: its cause is present since the birth, even if the nystagmus itself appears a little later on. It’s the contrary of the acquired nystagmus.

(22) **Oscillopsia**: permanent sensation of visual movement.

(23) **Actinic keratosis**: thickening of the skin provoked by an over-exposure to sunlight rays.

(24) **Carcinoma**: cancer that starts in the superficial tissues of the skin.

(25) **Basal cell**: the basal cell carcinoma is a cancer which starts in the basal layers of the skin just between the dermis and the epidermis.

(26) **Squamous cell**: the squamous cell carcinoma is a cancer which starts in the lay of the prickle cells of the skin within the epidermis.

(27) **Extended wear lenses**: the new materials used, permeable to oxygen allow to wear this type of lenses day and night up to 30 consecutive days.
"Genespoir supports my view that I’m not alone and that we represent a force for stepping forward together."

Bernard, cartoonist and musician.