ENTIDAD DECLARADA DE UTILIDAD PÚBLICA

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AHUCE

ASOCIACIÓN HUESOS DE CRISTAL DE ESPAÑA
OSTEOGÉNESIS IMPERFECTA

Contact

Ahuce

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AHUCE and OI

AHUCE stands for the National Brittle Bones Association of Spain, a public service organization that was founded in 1994 with the purpose of addressing the specific needs of people with Osteogenesis Imperfecta and their families.

What is Osteogenesis Imperfecta?

Osteogenesis Imperfecta (OI) is a genetic disorder that in most cases is caused by a decrease in the quality and quantity of collagen. This shortage (and in other cases, the lack of certain enzymes) causes the bones of people with OI to be fragile and to fracture easily.

The primary symptom of people with Osteogenesis Imperfecta is the frequent occurrence of bone fractures and ligament lesions. The level of intensity varies greatly. Other clinical manifestations of OI include:

- Bone fragility and deformity
- Muscle weakness and lax articulations
- General muscle and bone pain
- Progressive deafness
- Scoliosis
- Short stature
- Brittle Teeth (DI)

All symptoms are not found in every person with OI. There is a great variety of genotypes and phenotypes, so the intensity varies from mild to grave.

Osteogenesis Imperfecta is a rare disease because its prevalence is somewhere between 1:10,000 and 1:15,000 of births. According to these stats, we can estimate that between 3,000 and 4,500 people have OI in Spain. In the more mild cases, the disorder can remain unknown to doctors that treat the patient.

The diagnostic of a person with OI continues to be clinical, given that the great genetic diversity of the impedes/hampers the obtainment of genetic confirmation. Although in some cases it is possible to detect characteristic signs of OI in an ultrasound around the sixteenth week of gestation, there are people with OI who take years to be diagnosed. And there are cases where they are not diagnosed until adulthood.

The classification of OI is not easy, since the clinical characteristics vary greatly, and a single genetic type of OI varies greatly from person to person.

The actual classification of the distinct types of OI is based on the traditional classifications (established by an Australian doctor, Dr. Sillence, in 1970) and recent genetic discoveries which have added new types. There now are the following types of OI:

- **Type I:** The most common type of OI. Within type I there is a great variety of manifestations, from people who suffer only a few fractures in their life to people with frequent and disabling fractures and lesions. Those with type I have average stature.

- **Type II:** Characterized by a short stature and grave deformities. Although present treatments have improved considerably the outlook for these patients, some children born with type II pass away after only a few hours of life due to respiratory problems.

- **Type III:** A type of OI with variable prognostic thanks to present treatments. Those with type III OI have short stature. This type of OI usually is accompanied by great fragility and curves in the spine and extremeties.

- **Type IV:** An extremely variable type of OI, with some mild manifestations and others that are moderate or grave. Those with type IV OI usually have a slightly shorter stature than the norm.

- **Type V:** Similar to type IV, but with the unique formation of hypertrophic calluses at fracture sites.

- **Type VI:** This type of OI is not caused by a change in collagen levels, but by an error in the synthesis of a protein. This is a rare form of OI with a recessive mode of inheritance that is caused by a mutation in the gene SERPINF1. It is distinguished by a defect in the mineralization of bone.

- **Type VII:** In appearance and symptoms type VII is very similar to type V and II. Clinical characteristics include short stature, shortening of femurs and humerus. This is a recessive type of OI that is caused by a mutation in the gene CRTAP.

- **Type VIII:** Clinically type VIII looks like types II and III in appearance and symptoms. It is characterized by a grave growth deficiency and extreme demineralization of the skeleton. This is a recessive type of OI that is caused by a mutation in the gene LEPRE1.

- **Type IX:** This is a recessive type of OI with variable clinical manifestations from moderate to lethal. It is caused by a mutation in the gene PPIB.

- **Type X:** This is a recessive type of OI with a clinical manifestation that varies between grave and lethal. It is caused by a mutation in the gene SERPINH1.

- **Type XI:** This is a progressive type of OI that can be characterized by spine deformations and is also called Bruck syndrome. It is a recessive type of OI and is caused by a mutation in the gene FKBP10.

Advances in genetic research are leading to important discoveries in this field. Likely new types of Osteogenesis Imperfecta will be added to this list in the future.

OI is a chronic disease with no cure. Nevertheless, advanced therapeutics can greatly improve the quality of life of those with OI. Three therapeutic pillars form the basis of treatment for OI:

- Physical Therapy: with specific physical therapy treatment fractures are prevented, posture defects are corrected, and bones and muscles are strengthened.

- Pharmaceutical: biphosphonates and other drugs affect the bone metabolism and successfully decrease the number of fractures.

- Surgery: with interventions and insertion of surgical material bone alignment can be maintained and deformities fixed.

As a complement to these three pillars, other therapies have also proven successful for the treatment of OI, including dentistry, ophthalmology, ENT work, and psychological help.

Objectives of AHUCE

The objectives AHUCE are:

• Serve as the link between partners and defend the interests of people with OI.

• Publish medical, physical therapy, social, educative, and all other information that would be of interest and that contributes to bettering the quality of life of those with OI.

• Support and manage networks of OI professionals as a resource for those affected with OI.

• Support research in the field of OI.

• Fight for the right to education, work, and health, the economic benefits coverage, and other defined services in the current laws. In this way we fight for a more accessible society in terms of the architectural sphere, transportation, and media.

• Work towards the integration of and against the discrimination of people with OI into schools and jobs.

• Collaborate with other organizations of people with disabilities, rare disease, or OI in Spain and the world.

• Form relations between OI organizations and the research, medical, and health professionals and organizations related to OI.

• Create resources that allow for a better social adaptation and integration of people with OI.

AHUCE belongs to the following organizations:

- **FEDER:** Spanish Federation of Rare Diseases.
- **COCEMFE:** Spanish Confederation of Personals with Physical and Organic Disabilities.
- **OIFE:** Osteogenesis Imperfecta Federation Europe.
- **EURORDIS:** European Rare Diseases Organization.

AHUCE is also closely connected to the **AHUCE Foundation**, an organization created from the association, whose objectives are to promote research in the field of Osteogenesis Imperfecta and develop International Cooperation activities related to Osteogenesis Imperfecta. Through AHUCE Foundation, the association is also in constant contact with entities, associations, and foundations of Osteogenesis Imperfecta in Europe, America, Africa, and Asia, and belongs to **ICORD**, the International Conference for Rare Diseases. The AHUCE Foundation works with Ahuce to start cooperation projects, research, and the training of professionals dedicated to Osteogenesis Imperfecta.

Services AHUCE provides

Information and General Counseling

• Information and Orientation Service (SIO), which can be provided face-to-face or over the phone, gives direct support to those affect with OI and their families.
• Publication Service, which provides educative and informative material about the most recent news in OI for those affected and for professionals in the public health sphere.

Physical Therapy

• Physical Therapy Second Opinion and Attention Service: physical therapy can be done in office or at home, and consultations are over the phone or email.

• Physical Therapy Training for Professionals. This includes a consultation service and physical therapy forum and network.

Psychology

• Psychological services for the affected and their families.

Hospital Accompaniment and Rest for the Family

• Carried out by different professionals and volunteers. Support and advice is offered during hospital visits for consultation, check-up, surgical or pharmaceutical treatment.

Orthopedic Surgery

• Orthopedic Surgery Second Opinion Service: consultations in the AHUCE office, over the phone, or through email. Collaboration with Dr. Parra and Dr. Bueno of Hospital Universitario de Getafe.

Dentistry

• Dentistry Second Opinion Service is offered by Dr. de Nova in the Dentistry department of the Complutense University of Madrid.

School Counseling

• Individual teachers receive counseling and support to manage the specific needs of a child affected with OI in school, with the goal of promoting the integration and success of the child.

• Specific preparation for physical education teachers to help them adapt their curriculum for children with OI.

Agreements and additional benefits for the members

• Collaboration with the Upper Institute of Shiatsu Yasuragi. This is a free service for members of AHUCE.
• Agreement with CEORTEC, Orthopedic Technology of Pozuelo (Madrid), for buying orthopedic prosthetics and technical aids for a reduced price for members of Ahuce.

• Collaboration agreement with the genetic companies Sistemas Genomics (Valencia) and CAGT (Zaragoza) for reduced prices of genetic screenings of embryos for members of Ahuce.
• Collaboration Agreement with TACOVAL, a company dedicated to the adaptation of cars for people with functional difficulties, for a reduced price for members of Ahuce.

Ahuce's Activities

Annual Members Conference

Once a year during a weekend people with OI and their families from all over Spain get together. Talks about medical news and a variety of workshops are given according to the necessities and desires of the group. Examples of workshops include physical therapy, psychology, school, and primary care. Fun activities are also planned.

National OI Convention

A three-day convention between Doctors and Patients is held annually and attended by health professionals with experience in the treatment of people with Osteogenesis Imperfecta. The purpose of the convention is to share the most recent medical information about treatments, diagnostics, and other questions relevant to Osteogenesis Imperfecta. In this way Ahuce puts patients and specialists in contact within our country and around the world. The environment of the convention also encourages the formation of personal relationships between patients, their families, and health professionals. The convention also offers workshops and fun activities for the attendees.

Medical and Physical Therapy Convention

Along with the annual convention, every other year Ahuce organizes day conventions for highly specialized training for medical and physical therapy professionals. The speakers at the day convention are internationally renowned Osteogenesis Imperfecta specialists.

Attendance at International Events

Aware of the importance of having a relationship with the international community of OI, and with the purpose of keeping our members updated on the new therapies and discoveries related to OI, Ahuce maintains its relationships with international entities and regularly attends world conferences and reunions, and is noted for its continual support. Only in the past five years has the Ahuce association and foundation been present as participants and speakers at the annual reunions of OIFE, which occur in different countries in Europe, and at the the International Conference of OIFE about physical therapy in OI (Germany, 2009), the Second Osteogenesis Imperfecta Latin American Conference (Ecuador, 2011), the International Conference of OIFE about social aspects of Osteogenesis Imperfecta (Portugal, 2012), the First Osteogenesis Imperfecta Day Conference of Central America (Panama, 2013), and the ICORD International Conference (Russia, 2013). International relationships offer Ahuce a unique opportunity to receive and offer information about the most recent clinical advances in OI.

Informative Activities

Ahuce offers information about the news and activities related to OI in many different forms:

• **Webpage.** Ahuce maintains an updated webpage that is a source of corporate, medical, and social information for its members. www.ahuce.org and www.fundacion.ahuce.org

• **Social media.** The association has a Facebook (www.facebook.com/Osteogenesis.imperfecta.Ahuce) and a Twitter (www.twitter.com/AHUCE)

• **Ahuce Magazine.** Ahuce produces a periodic magazine in which it publishes medical talks, articles relevant to OI or disabilities in general, new laws, articles of our members and friends, and corporate information direct from the office and board:
www.ahuce.org/PublicacionesAHUCE/RevistaVocesdeCristal.aspx

• **Periodical Newsletter.** This was resumed recently to quickly and concisely divulge information and news about laws, the company, courses, and calls.
www.ahuce.org/PublicacionesAHUCE/Boletin90dias.aspx

• **Activities to increase global awareness.** Ahuce participates in commemorative and awareness activities. Notably Ahuce participates in Rare Disease Day on February 28th and Osteogenesis Imperfecta Day on May 6th.

• **OI Pamphlets.** In collaboration with the Ahuce Foundation, Ahuce edits and publishes a series of pamphlets about various clinical and medical aspects of Osteogenesis Imperfecta. The pamphlets of Osteogenesis Imperfecta are free and you can read or download them directly at www.ahuce.org/PublicacionesAHUCE/BoletinesOI.aspx

• Through their webpage, Ahuce also makes available to the medical community the most recent news about OI from books and articles.

• Ahuce's presence in press, radio, and television are also available in communication section of the webpage www.ahuce.org/Prensa/TelevisionyRadio.aspx

A small donation goes a long way

Ahuce is a Non-profit organization that depends on private donations and grants from public and private organizations.

To continue working and contributing to bettering the conditions and the quality of life of persons with Osteogenesis Imperfecta we need financial help. As AHUCE is a public service organization, all the financial support given by individuals to AHUCE can lead to tax exemption in Spain.

With your donation, we can collaborate in projects, and contribute to the execution of numerous activities that are already in progress. With you donation, no matter how small, you contribute to the well-being and social inclusion of children and adults that live with Osteogenesis Imperfecta. There is still so much to be done.

Account for donations:

Title: AHUCE, Asociación Nacional Huesos de Cristal

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Important: To get tax exemption, send us a message with the receipt, your name, and NIF (DNI, NIF, or NIE).

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Osteogenesis imperfecta

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