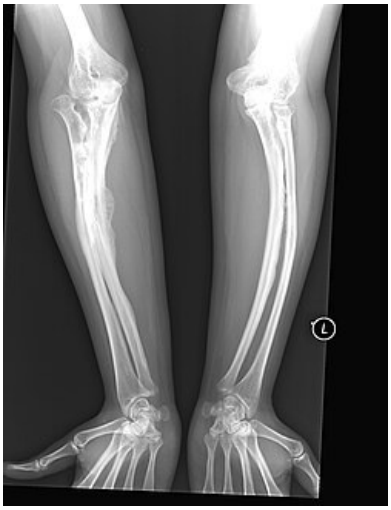
Osteogenesis imperfecta

Historical considerations

The Osteogenesis Imperfecta (OI) was identified as a disease in the 17th century, however only in the 19th century was it recognized by the current designation. The system used today was presented by Sillence *et al*, in 1979, which subdivides the disease in 4 groups depending on the severity and progression.

Characterization of disease



Osteogenesis Imperfecta is a hereditary disease caused by mutations in genes coding for collagen, which leads to the formation of very fragile bones. Mutations that underlie this disease are transmitted to offspring in a autosomal dominant inheritance (85-90% of mutations). However, they are currently described 8 different types of this disease two of which (types VII and VIII) are inherited as autosomal recessive form and do not include mutations in genes coding for collagen

Prevalence and symptoms

This genetic disorder affects between 6 to 7 individuals in 100.000 worldwide. Types I and IV are the most common forms of OI and affect 4 to 5 Individuals in 100000. Symptoms vary from person to person, even among individuals with the same type or within the same family. The main symptoms are blue scleras, dentinogenesis imperfecta, bone fragility, fragile ligaments and deafness in the case of the adult.

Different types of OI

8 different types are determined by the severity and progression of the disease:

Type	Heredity	Severity	Fractures	Stature	Sclerotic	Deafness	Mutated Gene
I	AD	Light	Less than 100	Normal	Blue	Yes (50%)	COL1A1
II	AD	Very Severe	Multiple	Very Short	Dark Blue	-	COL1A1 or COL1A2
III	AD	Severe	Multiple	Very Short	Blue	Yes	COL1A1 or COL1A2
IV	AD	Moderate to Light	Multiple	Variable	White	Partial	COL1A1 or COL1A2
V	AD	Moderate	Multiple	Variable	White	No	-
VI	AD	Moderate	Multiple	Short	White	No	-
VII	AR	Moderate	Multiple	Short	White	No	CRTAP
VIII	AR	Severe	Multiple	Very Short	White	Yes	LEPRE1

AD - autosomal dominant; AR- Autosomal recessive

Diagnosis

The first diagnostic approach is the study of differential family history and observation of patients for the identification of phenotypic changes, in the latter case it may be necessary to use x-rays to check the degree and type of fractures. For a more specific diagnosis, other types of tests such as skin biopsy are performed for analysis of structure and quantity of collagen. To confirm the differential diagnosis and prenatal diagnosis molecular tests have been carried out to detect the mutation and therefore be able to provide appropriate treatment to each patient.

Treatment

The Osteogenesis Imperfecta is a disease that has no cure, however, these patients should practice some types of exercise and using, when appropriate, walking aids with the aim of improving their quality of life. In the most serious cases it is necessary to resort to surgeries and hormone treatments. The therapy that now offers a higher rate of success is the use of bisphosphonates. Although not a cure for this disease has the ability to increase bone mass and consequently reduce the number of fractures.

Future prospects

The therapy used is currently restricted to the use of orthopedic equipment and bisphosphonates, both with limited success. The use of bisphosphonates improves patients' quality of life, however is not a cure for this disease. Future prospects are directed to the use of gene therapy or cell therapy.

Links

- STEINER, Robert D – PEPIN, Melanie G – BYERS, Peter H. *Osteogenesis Imperfecta* [online] . 2nd edition. 2005. Available from <http://www.ncbi.nlm.nih.gov/books/NBK1295/>. ISBN 20301472.
- GENETICS, Home References. *Osteogenesis imperfecta* [online]. The last revision November 2007, [cit. 2012-12-04]. <http://ghr.nlm.nih.gov/condition=osteogenesisimperfecta>.

Related Articles

- ROUGHLEY, PJ. , et al. Osteogenesis Imperfecta-Clinical and Molecular Diversity. *European Cells and Materials.* 2003, y. 2003, vol. 5, p. 41-47, ISSN 14562771.
- GLORIEUX, FH. , et al. Experience With Bisphosphonates in Osteogenesis Imperfecta. *Pediatrics.* 2007, y. 2007, vol. 119, p. 163-5, ISSN 17332237.

Other References

- National Institute of Health (NIH). *Osteogenesis Imperfecta Overview* [online]. [cit. 2012-12-04]. <http://www.niams.nih.gov/Health_Info/Bone/Osteogenesis_Imperfecta/overview.asp>.