

Frequently Asked Questions

1. If no members of my family are affected by OI, can I still have a child with the condition?

Yes. Most infants with severe forms of OI are born into families with no history of OI. The cause of a spontaneous mutation occurring in an individual is not known and is not due to anything that parents did or did not do during or before pregnancy.

2. If I have OI, will I definitely pass on the condition to my child?

No. For the majority of cases, there is a 50% chance with each pregnancy of having an affected child. A small number of OI are inherited in an autosomal recessive pattern, in this case there is a 25% chance of having a child with the condition if both parents are carriers of the mutation.

3. If I have one affected child, does that mean I will not have another?

No. The risk depends on the mode of inheritance for the child's OI. Genetic counselling with a genetic counsellor will clarify this issue.

4. Will increasing calcium intake help in strengthening the bones of a person with OI?

Generally, the diet has nothing to do with OI. The correct amount of nutrition should be advised by a doctor as excessive amounts may cause side effects.

5. Are the fractures associated with OI unique and easily distinguishable from other fractures?

No. The fractures are similar in appearance hence resulting in the rare occasions where the question of child abuse is raised when the child is first seen in a hospital.

6. Would a child with OI be able to go to school?

Yes. Precautions need to be taken to prevent knocks and falls which could lead to fractures. Many with the condition have gone on to show excellent academic achievements.

References

Facts About Osteogenesis Imperfecta [online]
Available from: http://www.oif.org/site/PageServer?pagename=AOI_Facts [Accessed March 2010]

Genetics Home Reference [online]
Available from: <http://ghr.nlm.nih.gov/condition=osteogenesisimperfecta> [Accessed March 2010]

NIAMS Health Information [online]
Available from: http://www.niams.nih.gov/Health_Info/Bone/Osteogenesis_Imperfecta/default.asp [Accessed: March 2010]

For further Information, please contact us at:

**Persatuan Penyakit Jarang Jumpa Malaysia
(Malaysian Rare Disorders Society)**

16, Lorong 5/10D
46 000 Petaling Jaya
Selangor Darul Ehsan

Telephone: 019-382 7386/019-771 4543

Fax: 03-7958 8459

E-mail: info@mrds.org.my Website: www.mrds.org.my

© December 2010 Malaysian Rare Disorders Society



Rare Disorders Series:
**Osteogenesis
Imperfecta (OI)**

What is Osteogenesis Imperfecta (OI)?

OI is a group of genetic disorders characterized by bones which break easily from mild trauma or with no apparent cause.

There are eight types of OI. Each type is different in its characteristics and severity.

Range of symptoms	Type of OI
Mild	1
Moderate	4, 5, 6
Severe	2, 3, 7, 8

Four genes have been associated with causing OI. Genes are located on chromosomes and contain 'recipes' to make proteins.

90% of OI cases are caused by mutations in the *COL1A1* or *COL1A2* genes. These genes provide instructions for making proteins that are used to assemble type 1 collagen.

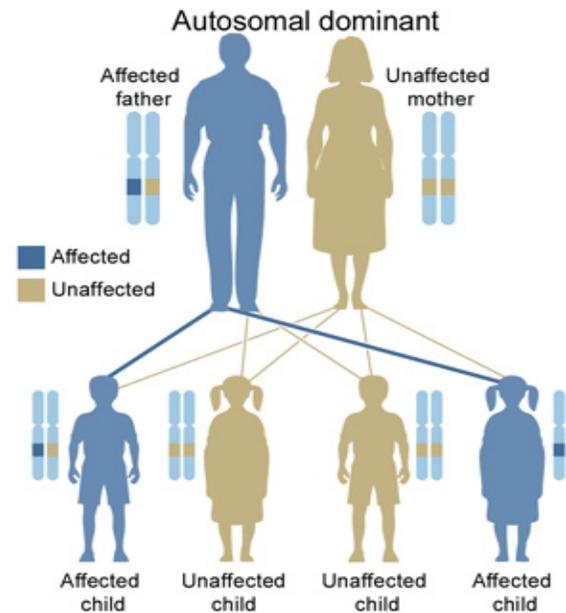
This type of collagen is the most abundant protein in bone, skin and other connective tissues. Reduced amounts or altered forms of type 1 collagen cause brittle bones to fracture easily.

OI may be inherited or may occur spontaneously (the first in the family) in an individual.

Most types of OI have an autosomal dominant type of inheritance. This means, only one copy of the altered gene is required to cause the condition.

The prognosis for an individual with OI varies greatly according to the severity of symptoms.

Patients are advised to consult a geneticist to obtain further information on disease progression.



U.S. National Library of Medicine

*Autosomal dominant pattern of inheritance
in majority of patients with OI*

Signs and symptoms

- Multiple bone fractures
- Malformed bones
- Blue sclera (the white part of the eye)
- Brittle teeth (Dentinogenesis Imperfecta)
- Hearing loss (starting at age 20s or 30s)

Other ways OI may affect the patient

- Nutrition related problems i.e. constipation, difficulties eating solid food, failure to thrive, malnutrition and obesity
- Respiratory problems due to rib fractures
- Fatigue
- Chest and back deformities (scoliosis)
- Psychosocial difficulties

Note that symptoms and severity vary according to type of OI and not all of the symptoms stated above will be seen in an individual with OI.

Testing

Genetic counselling by qualified personnel is advised if there is any family history of the condition and before any genetic testing is done.

- Fetal Ultrasound
To check for skeletal fractures and limb deformities in the foetus.
- X-ray
To check for skeletal fractures and deformities in infants and children. Milder forms may be missed.
- Genetic Testing
To confirm and diagnose the type of OI affecting an individual by looking at the mutation.

Treatment

There is currently no known cure for OI.

Treatment is focused on minimizing fractures, maximizing mobility, independent function and general health.

Pamidronate, a type of drug which increases bone density and regulates bone formation has thus far shown considerable success in the treatment of severe OI.

Safe exercises such as swimming are encouraged to promote a healthy lifestyle.

Orthopaedic surgery to implant rods may be recommended to increase support to bones.

Mobility aids such as canes, braces and wheelchairs may increase independence and the ability to care for oneself.