



Adults with Osteogenesis Imperfecta

Osteogenesis Imperfecta (OI) is a genetic disorder characterised by bones that break easily, often from little or no apparent trauma. It is caused by a genetic defect that affects the body's production of type I collagen, which is the major protein in connective tissues such as bone, ligaments, tendons and skin. This means that OI often affects individuals beyond the bones.

OI exhibits wide variations in appearance and severity, so a classification system has been identified to describe the different types of OI. Severity can also be described as mild, moderate, or severe.

Because OI is a highly variable disorder, it results in different physical features and clinical symptoms from one person to another. Therefore, people with OI should discuss these issues with their doctor to determine their individual risk and best course of treatment. Adults with OI need to cope with the musculoskeletal (e.g. bones, joints, ligaments, tendons) concerns associated with OI and also manage all of the same health issues as other adults.

This fact sheet provides a brief overview of health issues of concern to adults with OI, and should be read in conjunction with our factsheet about Osteogenesis Imperfecta.

Most adults with OI experience a decrease in the rate of broken bones (fractures) after puberty. Other medical problems, some of which are related to the basic genetic defect that cause OI, may require more attention. Tendon, muscle, and joint problems may be aggravated with time, and hearing loss may become significant. Regular medical review remains as important as it was during childhood.



The Health Care Team

An important part of managing OI and staying healthy is having a good relationship with your GP and medical specialists. The consultant who looks after OI in your area may be an endocrinologist (hormone specialist) or a rheumatologist (joint specialist). Your GP or hospital consultant may recommend advice from other specialists such as orthopaedic surgeons, or neurologists if required. You may also need input from occupational therapists and physiotherapists.

It will help your doctors and health care team if you are able to give them accurate information about your medical history and medication. If you are unsure about anything, don't be frightened to ask if you need to have things explained more than once. It's important that you understand what your health care team are telling you so you can be involved in decisions and look after yourself well.

Health Concerns Related to OI

Fractures and Medication: The frequency of fractures often decreases when people with OI reach adulthood. If you have received treatment with bisphosphonates as a child you may be able to have some time off treatment or stop the treatment. When to stop treatment may be different for different people, and your specialist will discuss this with you.

Bone Density: Bone density measurements are performed in adults with OI and we are working on a guideline to understand when and how often it should be done. It is important to note that if you have OI a low bone density does not mean you have osteoporosis. This is important as the treatments for osteoporosis may not work in OI. We recognise many patients and doctors get these two conditions confused, but they are different and patients with OI do not have osteoporosis.

The most widely used bone mineral density (BMD) test is called a dual energy x-ray absorptiometry (DXA) scan. It is painless: a bit like having an x-ray, but with much less exposure to radiation. It can measure bone density at the hip and spine. If possible, DXA scans should be done using the same machine on each occasion to avoid variations in test results caused by different equipment. DXA machines can also be used to take an image of the whole spine to look for fractures and curvature (vertebral morphometry).

Bone density can decline as a direct result of OI, from immobilisation due to limited weight bearing activity (for example after fractures), and from age-related changes in hormones and bone.

Smoking, overuse of alcohol, poor dietary calcium intake or low vitamin D, and some medications, including steroids, can reduce bone density. Your health care team can advise you on things you can do to maintain your bone density.



Treatments for low bone density can include calcium and vitamin D supplements and medication including oral or intravenous bisphosphonates, although in adults we are not sure if they work.

Muscle and joint problems: Adults often report pain in their lower back and hips. This can be the result of fractures of the spine, scoliosis (curvature of the spine), or joint deterioration. Other problems can include fractures that have failed to heal (non-union fractures) or differences in leg length (LLD). Muscle weakness, knee pain, ankle instability, and rolling in of the ankle are often a problem for people with OI. These problems can be a result of joint laxity or excessive joint flexibility.

Physiotherapy, orthotic devices or braces to improve hip, knee and ankle alignment are often helpful. Heel lifts and firm ankle supports can limit wear and tear on the joints and improve walking comfort.



Sometimes spine or joint replacement surgery may be considered to correct more severe problems.

Chronic Pain: Chronic pain is defined as pain that persists or recurs for more than 3 months. The understanding of the causes of chronic pain in adults with OI is limited. Pain may come from old fractures, or unstable joints. It is important the type of pain is tested as different types of pain will respond to different types of painkillers. In addition to medication, it is important that patients are managed holistically and have checks of their lifestyle, mental wellbeing and physical fitness.

Breathing Problems: Breathing problems are the main concern of many OI adults, particularly those with OI Type III and Type IV with significant curvature of the spine. Decreased chest volume, chronic bronchitis, and asthma can lead to a reduction of lung capacity. Rib fractures and muscle weakness also may contribute to the problem.

Sleep apnoea is a related problem for some adults with OI and can be determined with an overnight sleep test. More information about sleep apnoea can be found here: www.nhs.uk/conditions/sleep-apnoea/

It is important that chest infections are treated quickly in OI. If you do develop a chest infection, you should see a doctor as soon as you can.

Basilar Impression (BI): This is a special problem for people with OI Type III and IV. BI involves pressure from the spinal column on the base of the skull. Symptoms can include headache, muscle weakness, and tingling or numbness of hands and feet. Evaluation by a neurologist, including MRI examination of the cervical spine and base of the skull, is necessary. A neurologist should monitor BI symptoms. It has been reported that some people have BI, but their symptoms do not get progressively worse.

Heart function: A small number of adults with OI may have heart valve problems. The most common is called mitral valve prolapse, in which one of the heart valves becomes weakened. Weakness of the main artery (aorta) may also occur but is not common.

High blood pressure (hypertension) is as common among adults with OI as in the rest of the population. High cholesterol and related cholesterol disorders that may occur in families can contribute to heart problems as well. Medical management of these disorders includes appropriate diet and drug therapies and regular monitoring by a primary-care doctor. Drugs such as statins can be very helpful along with diet in controlling lipid problems. Coronary artery surgery has been successfully performed on people with OI, although precautions are necessary because of tissue fragility.

Hearing: Approximately 50 per cent of all adults with OI will experience some degree of hearing loss during their lifetime. Hearing tests and MRI examination of the hearing canals can help the doctor understand the involvement of the bones of the ear. Treatment for hearing loss usually begins with hearing aids. Some adults are candidates for either stapedectomy or cochlear implant surgery.



Vision: The connective tissue problem in OI can extend to the eyes. Eye examinations are recommended every two to three years. OI can affect the shape of the lens and the strength of the coat of the eye, called the sclera. For this reason, adults with OI should consult with an ophthalmologist before using contact lenses. Also, laser lens surgery is not recommended for people with OI.

Teeth: People with OI often have fragile teeth (dentinogenesis imperfecta). It is important that you look after your teeth well with regular brushing and good dental hygiene and that you have regular dental check-ups.



Stomach: Stomach problems are not uncommon in OI. These include gastric acid reflux, and chronic constipation. These problems can usually be helped by medication.

Kidney Stones: There appears to be a risk of kidney stones in about 20 percent of people who have OI. These may be caused by changes in medications or in diet that increase the amount of calcium circulating in the body.

Starting a family: Women have to avoid getting pregnant while they are taking bisphosphonates, and you can talk to your specialist about options for contraception. If you are thinking about having children, you can discuss this with your specialist and early discussions before you are pregnant can be very helpful to make plans. You might wish to have advice from a geneticist about whether your children will have OI (with most forms of OI there is a 50% chance that a baby will inherit the condition).

There are reproductive options available for people with OI to have IVF and embryo selection on the NHS that you may wish to consider. You may also need to see a gynaecologist or obstetrician (pregnancy specialist) to discuss whether you might have any difficulties during pregnancy and delivery and what your options might be.



Weight: Maintaining a healthy weight is important. Being overweight not only increases risk for many health problems, such as diabetes and cardiovascular problems, but puts additional stress on joints.

Diet: OI adults with short stature may require less calcium and vitamin D supplements than usually prescribed. Total calcium intake of 800 to 1000 mg (milligrams) per day is usually sufficient. Supplemental vitamin D intake should not exceed 1000 IU/day unless levels have been checked and you have been advised on a higher dose by a doctor.



Physical Activity: Physical activity helps to maintain bone health, muscle strength and cardiorespiratory function. A low impact exercise program that may incorporate swimming, a stationary bicycle, or ski machine may be a good option for people with OI. It is important to exercise safely within the range of your abilities. Adults with OI may need to consult with a physiotherapist or rehabilitation professional to develop an individualised and appropriate program.



Alcohol Use: As for everyone, keeping alcohol intake at a low or moderate level helps to keep you healthy. People who are taking medications should check whether they might interact with alcohol. Impaired coordination resulting from too much alcohol or inappropriate mixing of alcohol and medications could lead to a serious injury for an adult with OI. Recommendations can be found here: www.nhs.uk/live-well/alcohol-support/calculating-alcohol-units/

Stress and Mental Health: Healthcare professionals also report that stress as the pressures of dealing with chronic health issues can put an individual at risk for mental health problems. Adults with OI, like other adults, should seek assistance if they experience anxiety or depression.



Tips for Staying Healthy as an Adult with OI:

- Maintain a healthy weight.
- Eat a balanced diet with appropriate levels of calcium and vitamin D.
- Do not smoke.
- Use alcohol only in moderation.
- Conduct a safe exercise program to maintain and develop muscle strength and aerobic fitness.
- Have regular medical check-ups as recommended for any adult.
- Test hearing every 2-3 years.
- Test vision every 2-3 years.
- Report any breathing problems.
- Seek help straight away if you have a chest infection.



Notes



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Compiled by the Brittle Bone Society in collaboration with BBS Medical Advisory Board and POINT (Paediatric Osteogenesis Imperfecta National Team). The information in this leaflet is correct as at 31st July 2021 but we cannot guarantee that it will be accurate and current at any given time. This leaflet is not intended in any way to replace the advice of your doctor or other medical professional. Leaflets are available online at www.brittlebone.org. This information is available in accessible formats on request.

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