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Paediatric Metabolic Bone Disease

There are approximately 206 bones in fully grown adults which provide both support and mobility and protect the body’s organs. They also help to control the calcium and mineral balance in the body. In childhood the ends of the bones are softer and made up of a tissue which is called cartilage, this is where the bones grow and during puberty this becomes solid. Each bone is composed of bone cells, cartilage (needed for growth), blood vessels and fatty tissue. Defects which are seen in bone disease can be caused by a number of factors including mineral abnormalities, such as calcium, phosphorus, magnesium and vitamin D which in turn can lead to bones which become brittle and fracture easily. Some specific conditions can also cause brittle bones, most notably Osteogenesis Imperfecta and Osteoporosis.

Features commonly seen in metabolic bone diseases include susceptibility to fractures even with minimal injury, abnormal bone growth such as bowing of the legs, bone and/or muscle pains and general weakness and lethargy.

Tests for metabolic bone disease often include blood and urine tests to measure levels of calcium, vitamin D and minerals. X-rays can show any deformities. Genetic testing is also now available. Bone density scans are available but may only be available in regional centres.

Vitamin D Deficiency

Vitamin D is needed by the body to absorb calcium in the gut; it also helps to strengthen the bones. It is a fat-soluble vitamin that is mostly produced in the skin upon exposure to sunlight. Vitamin D is formed when 7-Dehydrocholesterol is activated by UVB rays from the sun and it gets converted into more active forms in the liver and the kidney and is eventually turned into the chemical 1,25-dihydroxyvitamin D2 which is needed to maintain calcium balance in the body. Not all stores of vitamin D are produced in the skin from sun exposure; some of the stores come from the diet. However, approximately 90% comes from exposure to the sun and it is estimated that people require approximately 15 minutes per day for the body to have an adequate ability to synthesise vitamin D. People with darker skin may need double this time. This ultimately means that certain people are at higher risk of Vitamin D Deficiency, including those who have darker coloured skin, people who spend little time outdoors and people who do not expose themselves to sunlight due to the skin being covered for religious or cultural reasons. Poor diet may also contribute to Vitamin D Deficiency and those following a Vegan diet are at particular risk. Breast-fed babies whose mothers may have low levels of vitamin D are also at a higher risk.

Low levels of Vitamin D can cause bone pain, irritability and muscle weakness. During childhood, the bones may be soft and prone to bending or bowing of the limbs. There is also an increased risk of more serious conditions later in life such as heart disease and certain types of tumours.

Treatments of the condition includes children’s multivitamins which will provide the recommended daily dose for most vitamins. These are available from your GP or can be bought at a local pharmacy. However a doctor should be consulted first. If the vitamin D levels are very low, then higher doses will be needed and these may be available in the form of vitamin D injections. The disorder can be prevented in many cases by following a balanced diet that contains oily fish, beef liver, fortified margarines and cereals. The skin should also be regularly exposed to sunshine although it is important not to leave the skin prone to sunburn.

Primary Osteoporosis

Primary Osteoporosis is a genetic condition that is present from birth and is a disorder of collagen. Collagen is a protein which forms the framework for bone structure. In this condition there is a loss of bone mass that leads to a susceptibility to fractures. This is caused by the defect in collagen which means it can not support the mineral structure of the bones; making them more fragile.

Osteogenesis Imperfecta

Signs of Osteogenesis Imperfecta may vary greatly in severity. Symptoms include a susceptibility to frequent fractures with occur with minimal trauma, thin curved bones and crushed vertebral noticeable on x-ray, a blue tinge to the whites of the eyes, hearing problems, joint laxity and be prone to bruising easily. Individuals may also have discoloured or softened teeth which are prone to chipping, a short stature, an abnormal skull shape and heat intolerance or excessive sweating in the more severe cases. The more severe the case is the more deformities are present. There may also be a family history of the above features although symptoms may vary even amongst family members.

There are four different subtypes of this condition:

- **Type 1** is the most common type with symptoms including fragility, blue/grey whites of the eyes, mild motor delay, clumsiness and joint laxity. Individuals may tire easily. This is a milder form of the condition and there is no deformity.
- **Type 2** is a more severe form and babies tend not to survive more than a few months
- **Type 3** is characterised by severe progressive deformity
- **Type 4** is characterised by skeletal fragility and osteoporosis. Bowing of the legs is often seen.

Generally it is true to say that we all carry two copies of each gene. Osteogenesis Imperfecta may be passed down by an autosomal recessive mode of inheritance. A person who has one normal gene and one gene for the disease is termed a carrier for the disease and does not show any symptoms. The condition arises when an infant inherits a gene for the disease from both parents. The risk to the offspring of a couple who are both carriers is 25%. There is a 50% chance that their child will be a carrier. There is a 25% chance that the child will not carry the abnormal gene. This risk is the same for each pregnancy. The condition may also be inherited in an autosomal dominant pattern; this method of inheritance is when a single copy of the disease gene will dominate the other normal gene. Therefore, if a defective gene is inherited from either parent the child will be affected with the disorder. This means for each pregnancy if either of the parents has a defective gene, there is a 50% chance of a child being affected by the disease. These are general descriptions of the methods of inheritance, for further information a genetic counselling service should be consulted.

Treatment of Osteogenesis Imperfecta includes physiotherapy to help to strengthen muscles and improve mobility and the use of bisphosphonates which makes bone stronger by stopping re-absorption. These improve bone pain, mobility and reduce the susceptibility to fractures. In some cases surgery may be needed for individual fractures. In some cases rods may be inserted into the bones to strengthen them and protect against fractures.

Certain other groups of people are at risk of having a low bone density, this includes individuals who are on long term oral steroids and children with chronic inflammatory disease such as sever juvenile arthritis or Crohn’s disease. Those who have prolonged immobilisation perhaps due to another condition which causes weakness of the muscles, in turn leading to weak bones and children with Idiopathic Juvenile Osteoporosis are also susceptible.

This information is fully sourced and referenced, for more detailed information and references please contact CLIMB by email, letter or telephone.