

Cheme 355
In Class Exercise 1
11/6/02

name _____

We have discussed 3 diseases in Module #2, i.e. OI, Marfan Syndrome, and atherosclerotic cardiovascular disease. Use your notes from class and the following information sheet about Marfan Syndrome to answer the following questions.

- What is the etiology of OI?
- What tissue shows a majority of OI disease manifestations?
- Is OI communicable or non-communicable?
- Is OI a prenatal or post-natal disease?

Answer the same series of questions for atherosclerotic cardiovascular disease and for Marfan Syndrome.

Atherosclerotic cardiovascular disease

Marfan syndrome

Information about Marfan syndrome from <http://www.bsped.org.uk/NN/MARFAN.htm>

Cause:

Each human cell contains 23 pairs of chromosomes, one of each pair inherited from the mother and one from the father. Each chromosome carries many genes along its length and faults in genes are expressed as faults in the structure of body chemicals. Marfan syndrome is caused by a fault in a single dominant gene, this implies that the disease is present although only one of a pair of genes, either from the mother or the father, is affected. In about 25% of people with Marfan the fault in the gene has occurred spontaneously before birth (a mutation), in 75% it is inherited. Males and females are equally likely to be affected and, whether the gene was inherited or not, have a 50% chance of passing on the abnormal gene, and so the condition, to children of either sex. The gene for Marfan syndrome lies on chromosome 15 and carries the code for the structure of fibrillin which is an important part of collagen, the main component of connective tissue. Various different mutations of the fibrillin gene have been found to cause Marfan syndrome and this may account for the various different physical manifestations.

Features of Marfan syndrome:

People with Marfan syndrome are tall and usually of very slim build with especially long limbs, fingers and toes. They may have a high palate, either prominence or depression of the breast bone and sometimes curvature of the spine, especially in adolescence. They tend to have an early puberty and stop growing early so the final height of children is seldom as tall as seems likely through the school years. There is usually a degree of hypotonia (floppiness) of the joints, often with "double jointedness". These features may be apparent from birth or appear later in childhood. In the eyes the most common feature is short sightedness but there may be dislocation of the lens or a blue color of the white of the eye. The changes in the heart and blood vessels cause the most serious potential complications. The heart valves may be abnormally floppy and this can cause them to become leaky. There may also be widening of the aorta, the major vessel carrying blood from the heart to the body, and this can develop a tear in the wall (aortic dissection). Some children with Marfan syndrome have mild learning problems and there may also be a tendency to overactive behaviour.

Chem 355
In Class Exercise 2
11/6/02

name _____

Do you think gene therapy could be used to cure osteogenesis imperfecta?

Questions to consider:

Is the mutant gene in one location in the body or in every cell?

Is collagen made only in bone cells in the body?

Is it necessary to supply good collagen genes to all bone cells to cure OI?

Do you think gene therapy could be used to cure diabetes?

Questions to consider:

Is the mutant gene in one location in the body or in every cell?

Is insulin made only in pancreas cells in the body?

Is it necessary to supply good insulin genes to all pancreas cells?