



ML II AND ML III

Although originally thought to be two separate diseases, ML II and ML III are both caused by deficiencies of a targeting enzyme and are variations of the same disease. Individuals with more severe features have ML II; those with less severe, or attenuated, features have ML III.

What causes this disease?

The body constantly replaces used materials and breaks them down for disposal. This activity happens in the lysosomes of the cells. Enzymes responsible for breaking down the used materials can only reach the lysosome after a special signal has been attached to them. In ML II and ML III, the signal is not attached and the enzymes fail to reach the cell. The enzyme responsible for attaching the targeting signal is phospho-N- acetylglucosamine-transferase.

The cells filled with storage material are known as “inclusion cells,” hence the name “I- Cell” disorder. Babies may show little sign of the disease, but as cells sustain damage, symptoms start to appear.

How common are these diseases?

These diseases are very rare and sometimes misdiagnosed, so it is difficult to give accurate figures on frequency. The current estimate is that two or three individuals per 1 million births are diagnosed with ML II and ML III.

How is the disease inherited?

I-Cell and Pseudo-Hurler syndrome are caused by a recessive gene. There is a one in four chance with every pregnancy that the child will inherit the defective gene from each carrier parent and will be affected with the disease. There is a two in three chance that unaffected brothers and sisters of an individual with ML will be carriers.

Is there a cure?

At present there is treatment for symptoms as they arise but no cure for the underlying condition. Various experimental methods have been used to try to replace the missing enzyme, but none so far has been of any significant long term benefit.