Multiple Hereditary Exostoses

Presenting Symptoms

Benign bony growths or tumors on the skeletal structure which may result in limb disfigurement, pain, and/or mobility issues.

Other symptoms which may accompany Multiple Hereditary Exostoses:

- Uneven development of limbs
- Short Stature
- Cognitive/neurological difficulties.
  Discomfort and chronic pain, which may lead to fatigue and difficulty with concentration and focus.
  Significant mobility issues

About the Disorder

Multiple Hereditary Exostoses (MHE) is a disorder of the skeletal structure consisting of multiple bony growths, or exostoses, along the skeletal structure of the affected individual. MHE is known by a variety of names:

- Bessel-Hagen disease
- Diaphyseal Aclasis
- Exostoses, Multiple Hereditary
- Familial Exostoses
- Multiple Cartilaginous Exostoses
- Multiple Hereditary Exostoses
- Multiple Osteochondromas
- Multiple osteochondromatosis

Although some populations have higher incidences (such as the Chamorro population of Guam or the Ojibway population of Manitoba, Canada), the incidence of MHE in the general population is rare; thought to be about only one in 50,000 individuals. MHE is largely an inherited disorder; however, spontaneous mutations have not been that uncommon.

Historically, little has been known about this disorder, but research as of the past two decades has uncovered much. It has been determined that MHE is caused by a mutation in one of two genes, EXT-1 or EXT-2. This mutation results in abnormal levels of *heparin sulfate*, a long sugar-chain that helps bone cells grow and proliferate.

The manner of presentation of the disease may vary greatly from child to child. Some may only have a few tumors in varying areas of the body and go relatively unaffected by the disease. Others may have tumors in more problematic areas, suffer chronic pain, and have significant difficulties in terms of mobility and physical stamina. While tumors generally grow on the long bones of the skeletal structure (i.e., arms and legs), tumors may grow and present problems anywhere on the skeletal structure, from the finger bones, to hips, to ribs to bones along the neck, to even the skull.

Growths present on or near growth plates may result in stunted and/or uneven growth of one limb compared with another. This could result in significant fine and/or gross motor/mobility issues.

Recent research indicates that the same genetic dynamics which foster MHE may also contribute to brain-based social and emotional difficulties, such as Autism Spectrum Disorder. As thus, ASD and other brain-based disorders are present more frequently in children who have MHE.

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