

Hereditary multiple osteochondromas is inherited as an autosomal dominant genetic condition. Dominant genetic disorders occur when only a single copy of an abnormal gene is necessary to cause a particular disease. The abnormal gene can be inherited from either parent or can be the result of a new mutation in the affected individual. Approximately 10% of cases of HMO are thought to be the result of new mutations. At present two genes, EXT1 and EXT2, are known to show mutations in HMO patients and it is thought that these genes function as tumor suppressors. For some affected individuals no mutation in either gene is detected. In almost all these cases, the "mutation negative" patients do not have a familial history for exostoses. Most likely, they have an EXT1 or EXT2 mutation in only part of their body cells and the mutation is absent or undetectable in blood cells, which are usually used for DNA analysis. Data indicates that individuals with EXT1 mutations may have more severe effects than those with EXT2 mutations. The risk of passing the abnormal gene from affected parent to offspring is 50% for each pregnancy. The risk is the same for males and females. The prevalence of HMO has been estimated to be about 1 of 50,000 live births. A high prevalence of this disorder has been reported in some isolated communities. Hereditary multiple osteochondromas is a disorder that affects males and females in equal numbers but in general males tend to be more severely affected.