Homozygous OSMED (oto-spondylo-megaepiphyseal dysplasia) is an extremely rare genetic disorder characterized by malformation (dysplasia) of certain bones, hearing loss and distinct facial features. Skeletal malformations affect the bones of the arms, legs and spines eventually resulting in disproportionate short stature. Hearing loss is often severe. Intelligence is normal. Homozygous OSMED occurs because of disruptions or changes (mutations) to the COL11A2 gene and is inherited as an autosomal recessive trait. A diagnosis of homozygous OSMED is made based upon a thorough clinical evaluation, a detailed patient history, identification of characteristic symptoms, and a variety of specialized tests including x-rays. X-ray studies reveal characteristic skeletal malformations associated with homozygous OSMED. Genetic testing is also available to scan for mutations of genes coding for collagen XI.