Maxillofacial dysostosis is an extremely rare genetic disorder characterized by distinctive abnormalities of the head and face (craniofacial) area. Major symptoms include an underdeveloped (hypoplasia) upper jaw, downward-slanting palpebral fissures (which means that the opening between the eyelids slants downward), minor malformations of the external portion of the ears, and speech abnormalities. Maxillofacial dysostosis is inherited as an autosomal dominant trait. A second (distinct) form of maxillofacial dysostosis is believed to be inherited as an X-linked recessive trait. Maxillofacial dysostosis is inherited as an autosomal dominant trait. Genetic diseases are determined by the combination of genes for a particular trait that are on the chromosomes received from the father and the mother. Dominant genetic disorders occur when only a single copy of an abnormal gene is necessary for the appearance of the disease. The abnormal gene can be inherited from either parent, or can be the result of a new mutation (gene change) in the affected individual. The risk of passing the abnormal gene from affected parent to offspring is 50 percent for each pregnancy regardless of the sex of the resulting child. Maxillofacial dysostosis is extremely rare and the exact incidence of the disorder is unknown. Approximately 12 cases have been reported in the medical literature. Researchers believe that cases of maxillofacial dysostosis may go misdiagnosed or unrecognized making it difficult to determine the true frequency of the disorder in the general population. Maxillofacial dysostosis most likely affects males and females in equal numbers. A diagnosis of maxillofacial dysostosis is made based upon identification of characteristic symptoms, a detailed patient history, a thorough clinical evaluation and a variety of specialized tests to rule out other disorders.