

Orocraniodigital syndrome is an extremely rare inherited disorder characterized by multiple malformations of the head and face (craniofacial area) and the fingers and toes (digits). Major characteristics may include a vertical groove in the upper lip (cleft lip) and/or the inside, upper portion of the mouth (cleft palate), an abnormally small head (microcephaly), widely spaced eyes (ocular hypertelorism), improper development (hypoplasia) of the thumbs and/or toes, and/or webbing (syndactyly) of the toes. In some cases, malformations of certain skeletal bones may also be present. Mental retardation has occurred in the majority of cases. Orocraniodigital syndrome may be inherited as an autosomal recessive genetic trait. Approximately 10 cases of orocraniodigital syndrome have been reported in the medical literature. The symptoms are usually obvious at birth.