Norrie disease is a rare X-linked disorder that occurs due to errors or disruption (mutations) of the NDP gene. Norrie disease is a rare disorder that is fully expressed in males only. In rare cases, carrier females may display some symptoms of the disorder. The incidence and prevalence rates for Norrie disease are unknown. The disorder has been reported in all ethnic groups. A diagnosis of Norrie disease is suspected based upon a detailed patient history, a thorough clinical evaluation, and identification of characteristic findings. There may be a family history supporting X-linked inheritance. A diagnosis may be confirmed by molecular genetic testing in which a mutation in the defective gene (NDP) is identified. Such testing is available through clinical testing labs (www.genetests.org).