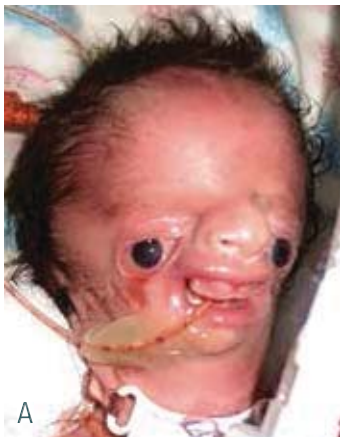

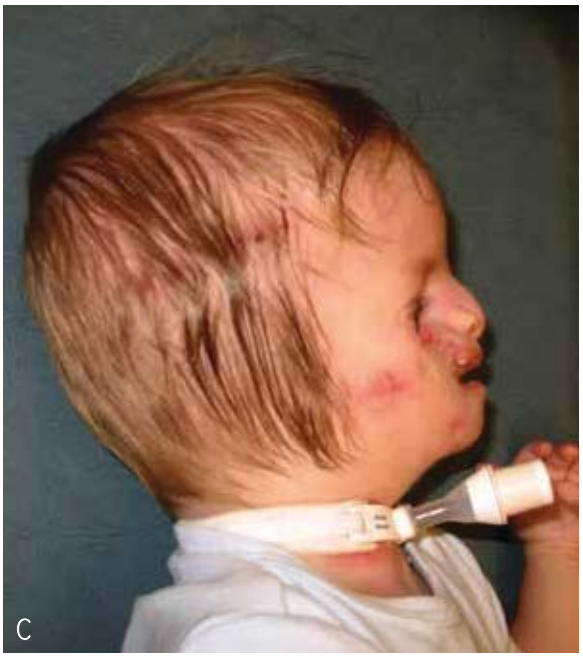

"It was really important to be able to tell the family why this abnormality happened, that it wasn't inherited and that it likely would not happen again in another child."

Individuals with acrofacial dysostosis, Cincinnati type, each have a heterozygous mutation in POLR1A, which encodes a core component of RNA polymerase 1. These images of an affected newborn show: (A) extensive craniofacial malformations at birth; (B and C) images taken at age 18 months after multiple reconstructive surgeries; (D) severe maxillary and zygomatic hypoplasia (black open-dashed arrow) and severe micrognathia and retrognathia (white block arrow); (E) severe microtia with absent pinnae (white arrows), external auditory atresia (white open-dashed arrows), and severe middle-ear hypoplasia and ossicular dysplasia (black open arrows); and (F) bilateral hip dysplasia and anterior bowing deformity of the femurs.