Acrodysostosis 1

National Cancer Institute

Source

National Cancer Institute. Acrodysostosis 1. NCI Thesaurus. Code C136464.

An autosomal dominant skeletal dysplasia caused by mutation(s) in the PRKAR1A gene, encoding cAMP-dependent protein kinase type I-alpha regulatory subunit. It is characterized by short stature, brachydactyly, and characteristic facial features. Resistance to multiple hormones is a common finding.