Immunodeficiency-Centromeric Instability-Facial Anomalies Syndrome 1

National Cancer Institute

Source

National Cancer Institute. Immunodeficiency-Centromeric Instability-Facial Anomalies Syndrome 1. NCI Thesaurus. Code C156430.

An autosomal recessive condition caused by mutation(s) in the DNMT3B gene, encoding DNA (cytosine-5)-methyltransferase 3B. It is characterized by immunoglobulin deficiency, centromeric instability of chromosomes 1,9, and 19 (rarely chromosome 2), and facial dysmorphism.