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ICF syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. ICF syndrome. ORPHA:2268

The Immunodeficiency, Centromeric region instability, Facial anomalies syndrome (ICF) is a rare autosomal recessive disease characterized by immunodeficiency, although B cells are present, and by characteristic rearrangements in the vicinity of the centromeres (the juxtacentromeric heterochromatin) of chromosomes 1 and 16 and sometimes 9.