1p36 Deletion Syndrome

National Cancer Institute

Source

National Cancer Institute. 1p36 Deletion Syndrome. NCI Thesaurus. Code C74983.

A rare syndrome caused by the deletion of the distal band on the short arm of chromosome 1. It is characterized by a distinctive facial appearance (microcephaly, deep set eyes, flat nose, and pointed chin), developmental abnormalities, mental retardation, seizures, hypotonia, hearing loss, and heart defects.