Open Peer Review on Qeios

Pseudotrisomy 13 Syndrome

National Cancer Institute

Source

National Cancer Institute. <u>Pseudotrisomy 13 Syndrome</u>. NCI Thesaurus. Code C125418.

A rare syndrome likely inherited in an autosomal recessive pattern. It is characterized by holoprosencephaly, polydactyly, phenotypic features reminiscent of trisomy 13, and normal karyotype.