IMAGe syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. IMAGe syndrome. ORPHA:85173

IMAGe syndrome is characterized by the association of Intrauterine growth retardation, Metaphyseal dysplasia (and short limbs), Adrenal hypoplasia congenita, and Genital anomalies. It has been described in less than 20 cases. The patients also present with dysmorphic features (frontal bossing, broad nasal bridge, low-set ears). In boys, genital anomalies include bilateral cryptorchidism, hypospadias, micropenis, and hypogonadotropic hypogonadism. This syndrome is likely to be transmitted as an autosomal recessive trait.