Dubowitz syndrome
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Published in Atlas Database: February 1998

Online updated version is available from: http://AtlasGeneticsOncology.org/Kprones/DUB10016.html
DOI: 10.4267/2042/37423

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Identity

Note: Dubowitz syndrome may be confused with Bloom syndrome; another differential diagnosis is fetal alcohol syndrome.

Inheritance: autosomal recessive; heterogeneity cannot be excluded; less than 150 cases described.

Clinics

Phenotype and clinics
- Phenotypic spectrum variable.
- Growth: from normal to severe retardation; intrauterine growth retardation is frequent; birth weight: 2.3 kg; length: 45 cm; cranial perimeter: 30 cm; delayed bone age.
- Head: microcephaly; high forehead; sparse hair; broad nose; epicanthus; hypertelorism; blepharophimosis; microretrognathia.
- Skin: eczema, a classical sign, may be absent.
- Congenital heart defects in 10%; other malformations: ocular, dental, skeletal, urogenital in male patients; frequent infections.
- Mental retardation in 30-70% of cases (from normal in 30% to severe retardation in 10%); seizures in 10% high-pitched voice; behaviour problems in 40%; most patients are 'hyperactive, shy, like music'.

Neoplastic risk
Haematological malignancies and pancytopenia in 10%, childhood myelodysplasia in particular; lymphomas.

Cytogenetics

Inborn condition
Appears to be normal or near to normal in most cases, although an increased rate of chromosomal breakage has also been described.

References


This article should be referenced as such: