Rubinstein-Taybi Syndrome

Main Features

- Mental Retardation
- Speech difficulties
- Broad thumbs and toes, often medially deviated

Eye Findings

- External:
  - Down-slanting palpebral fissures (88%)
  - Heavy eyebrows or high arched (76%)
  - long eyelashes (89%)
  - epicanthal folds (55%)
- Abnormal ERG (78%)
- Decreased Cone or Cone & Rod response
- Macular abnormalities (75%)
  - Hypoplasia, pigment abnormalities, increased red color, absent foveal reflex
- Abnormal VEP (60%)
- Strabismus (70%)
- Refractive Error (50%)
- NLD obstruction (40%)
- Ptosis (35%)
- Glaucoma (35%)
- Coloboma (25%)
- Congenital Cataract (25%)
- High Myopia (10%)
- Optic atrophy/disc abnormalities (10%)
- Chorioretinal Dystrophy (5%)
- Microphthalmia (5%)
- Nystagmus (4%)
- Ectopia Lentis (<1%)

Etiology

- 1/100,000 newborns
- Autosomal Dominant
- Many from Chromosome 16p13.3 microdeletions or mutation in gene for CREB binding protein found in this area (19%)
- Also 22q13: E1A binding protein

Other Findings

- Postnatal growth retardation
- Males 153 cm average
- Females 147 cm average
- Microcephaly
- Agenesis of Corpus Callosum
- Seizures
- Hypoplastic maxilla, micrognathia
- Beaked nose, deviated septum
- PDA, ASD, VSD
- Capillary Hemangiomas
- Sternal anomalies
- Hypospadias, Cryptorchidism
- Hirsutism
- Spina Bifida
- Syndactyly, Polydactyly
- Keloid formation

References

- OMIM #180849
- Wright, Spiegel eds. Pediatric Ophthalmology and Strabismsus 2nd ed. p 1051
- Ocular features in Rubinstein-Taybi syndrome: investigation of 24 patients and review of the literature. van Genderen MM et al. BJO 2000 Oct;84(10):1177-84 (photo)
- Rubinstein-Taybi syndrome: clinical features, genetic basis, diagnosis, and management

syndrome