Prader-Willi Syndrome

Main Features

- Neonatal
  - Neonatal hypotonia
  - Failure to thrive
  - Genital hypoplasia
  - Hypopigmentation of the skin, iris, and hair
- Childhood
  - Excessive eating and gradual development of morbid obesity
  - Delayed Motor milestones and language development
  - Cognitive impairment

Eye Findings

- decreased visual acuity
- iris and choroid hypopigmentation
- refractive error
  - Myopia 40%
  - Hyperopia 25%
  - Astigmatism 25%
- Amblyopia 16%
- Strabismus 40%
  - congenital ocular fibrosis syndrome
- cataracts
- diabetic retinopathy
- nystagmus
- congenital ectropion uvea
- Signs of oculocutaneous albinism can occur if deletion of the OCA2 gene which is found in the PWS critical region

Other Findings

- Short stature and growth hormone deficiency
- Delayed or incomplete secondary sexual characteristics
- Obesity complications
  - Sleep apnea
  - Diabetes mellitus
  - Atherosclerosis
- Epilepsy
- Scoliosis
- Behavioral problems and learning difficulties similar to autism spectrum disorder

- Scoliosis

Etiology

- loss of the paternal copy of chromosome region 15q11-q13

References

- EyeWiki Article
- Gene Reviews

syndrome