Angelman Syndrome

Main Features

- Severe developmental delay or intellectual disability
- Severe speech impairment
- Gait ataxia and/or tremulousness of the limbs
- Apparent happy demeanor that includes frequent laughing, smiling, and excitability

Eye Findings

- Strabismus
- Refractive error
- Deep-set eyes

Other Findings

- Seizures
- EEG shows large amplitude slow-spike waves

Etiology

- Deletion or defect in 15q11.2-q13 (80%)
  - Genes in this area control imprinting of other genes and several genes are imprinted in this area.
  - Imprinting leads to methylation or inactivation of certain genes so that material and paternal genes don't both function simultaneously.
- UBE3A mutation (11%)
  - Codes for ubiquitin ligase protein
  - Ubiquitination is a process of labeling proteins that targets them for degredation

Reference

- Gene Reviews
- Rare Disease Database Entry

syndrome