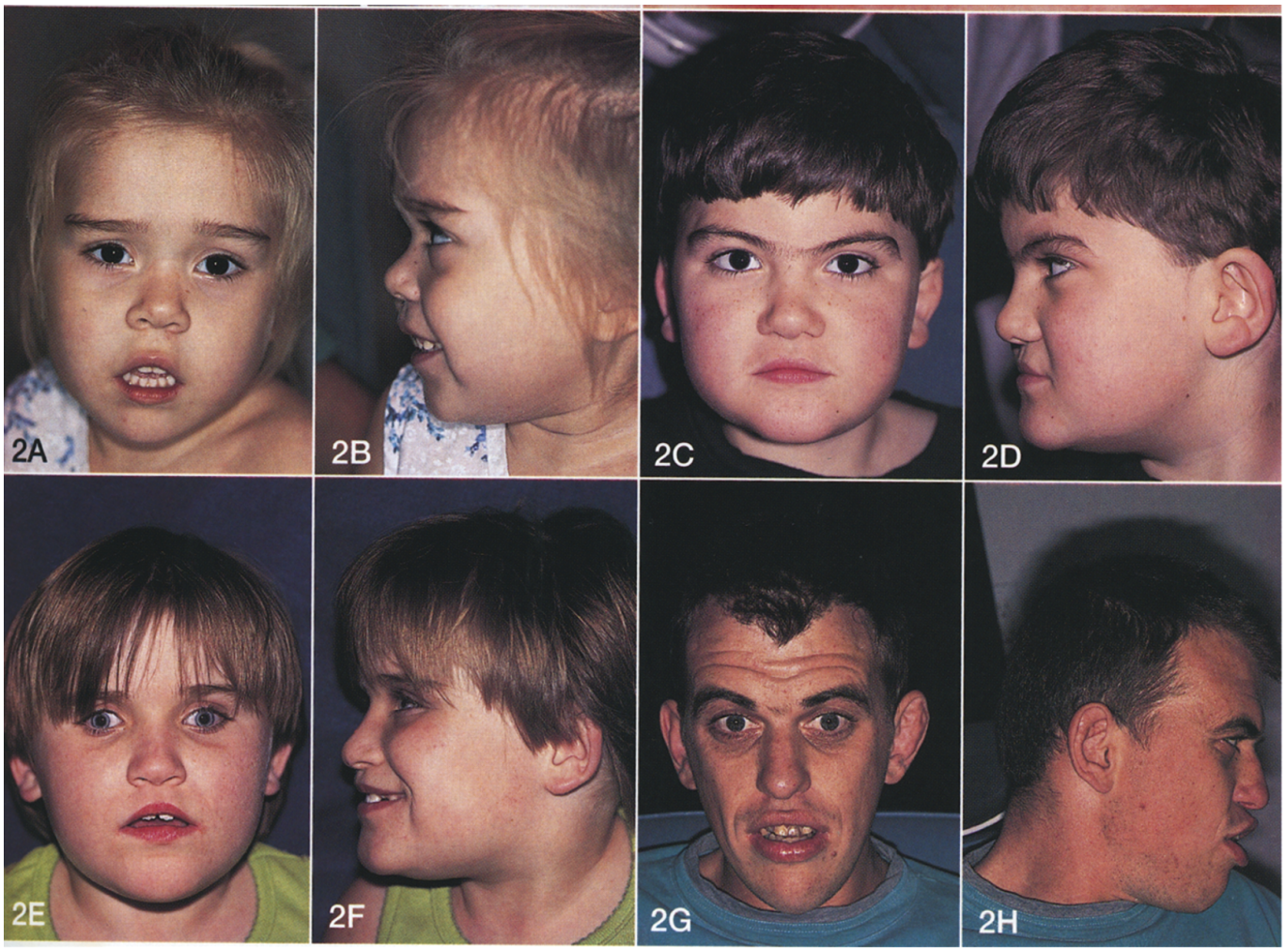
Smith-Magenis Syndrome

Main Features

- Distinctive physical features
  - Brachycephaly, broad nasal bridge, prominent forehead, synophrys, downturned upper lip, prognathism, malformed and malpositioned helices
  - Infants
  - School Age
  - Adolescent



From Ophthalmic Manifestations of Smith-Magenis Syndrome. Ophthalmology 1996

- Developmental delay
- Cognitive impairment
- Behavioral abnormalities
- Sleep disturbance
- Childhood-onset abdominal obesity

Eye Findings

- Iris anomalies (68%)
  - No iris collarette
  - Nasal correctopia
  - Stromal dysplasia
  - Mamillations
  - Wölfflin-Krückmann spots
- Microcornea (50%)
- Myopia (42%)
- Strabismus (32%)
  - Esotropia > Exotropia > Hypertropia
- Microphthalmos (rare)
- Uveal and Retinal Coloboma (rare)

Other Findings

- Clinical Features Table

Etiology

- Heterozygous deletion at chromosome 17p11.2 that includes RAI1 or a heterozygous intragenic RAI1 pathogenic variant.

References

1. Gene Reviews
2. Ophthalmic Manifestations of Smith-Magenis Syndrome. Ophthalmology 1996

syndrome