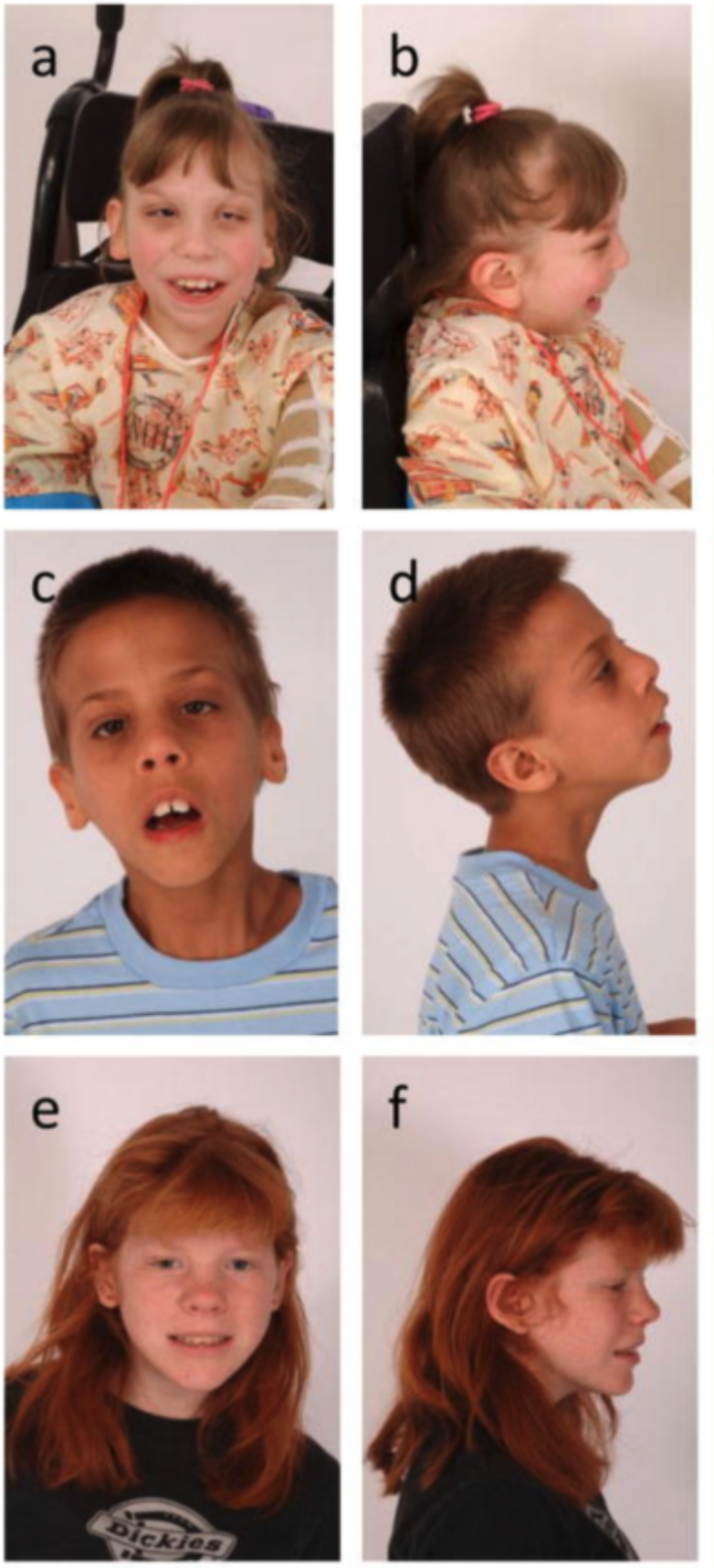
Smith-Lemli-Optiz Syndrome

Characteristics

- distinctive facial features
    - microcephaly with bitemporal narrowing, a short upturned nose with anteverted nares, unilateral or bilateral ptosis, epicanthus, and retrognathia



See Reference 1 below

- microcephaly
- intellectual disability or learning problems
- behavioral problems

    - autism
- Feeding difficulties and growth delay
- Malformations
    - heart
    - lungs
    - kidneys
    - gastrointestinal tract
    - genitals
    - polydactyly
    - syndactyly

Ocular Abnormalities

- Cataract (congenital or acquired)
- Strabismus
- Demylenation of optic nerves
- Sclerosis of lateral geniculate bodes
- Opsoclonus
- Poor visual following behavior

Etiology

- DHCR7 gene mutations
- Inherited in an autosomal recessive pattern.

References

1. Pathogenesis, Epidemiology, Diagnosis and Clinical Aspects of Smith-Lemli-Opitz Syndrome
2. NLM Link

syndrome