

Aase-Smith Syndrome

Main Features

Type I

- Severe joint contractures
- Hydrocephalus from Dandy Walker anomaly
- Overlap with [Gordon syndrome](#)

Type II

- Triphalangeal thumbs (finger-like)
- Anemia
- Might be the same as [Diamond-Blackfan Syndrome](#)

Eye Findings

- Downslanting palpebral fissures: euryblepharon, occasionally
- Ptosis
- Retinopathy occasionally

Other Findings

- Ventricular septal defect
- Cleft palate
- Congenital Neuroblastoma (1 case)

Etiology

- Most likely autosomal recessive
- Very few cases reported

Reference

- [OMIM](#)

[syndrome](#)

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