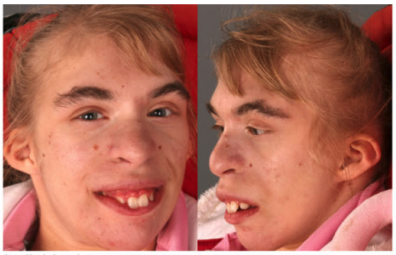
# Snijders Blok-Campeau Syndrome

## Main Features

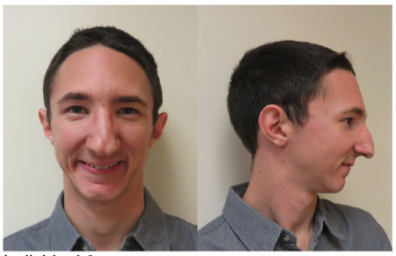
- Intellectual disability, speech problems and distinctive facial features


Individual 1


Individual 5


Individual 6


Individual 7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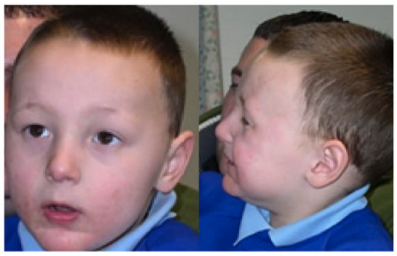
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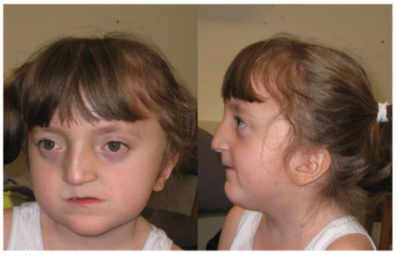
Individual 12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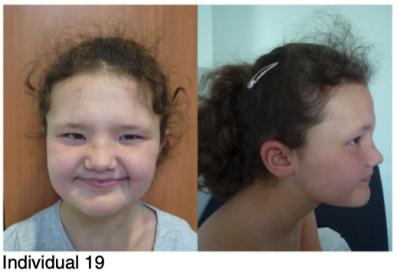
Individual 18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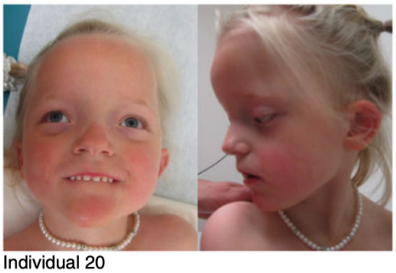
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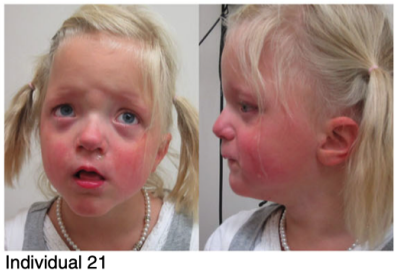
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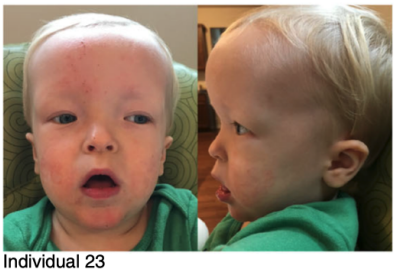
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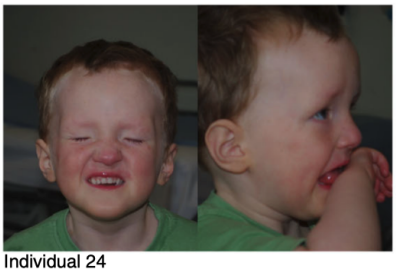
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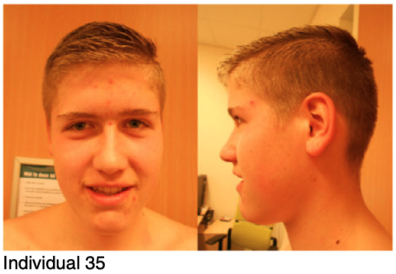
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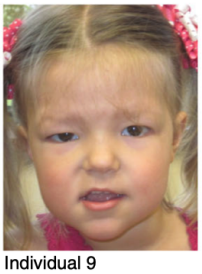
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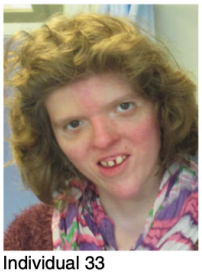
Individual 33


Individual 34

From Reference (1)

## Eye Findings

- Hypertelorism common
- Strabismus 30%

- CVI 9%

## Other Findings

- Macrocephaly
- Craniosynostosis
- Atrial Septal Defect
- Enlarged CSF spaces

## Etiology

- Mutations in the CHD3 gene which encodes for a protein that regulates gene activity through chromatin remodeling.

## References

1. Snijders Blok et al. CHD3 helicase domain mutations cause a neurodevelopment syndrome with macrocephaly and impaired speech and language. Nature Communications 2018;9:4619
2. Medline Plus entry

syndrome