SOX11 Syndrome And 2p25.2 Deletions

https://pubmed.ncbi.nlm.nih.gov/37924570/

Background:

SOX11 syndrome is a rare condition caused by deletions or de novo point mutations of the SOX11 gene. SOX11 is a transcription factor gene that plays an important role in brain development.

Aims:

The aim of this study was to quantitatively evaluate the behavioural profiles of individuals with SOX11 syndrome.

Methods and procedures:

The Vineland Adaptive Behaviour Scales 3 (VABS-3) and the Social Responsiveness Scale 2 (SRS-2) were completed by parents of 21 children and young adults with SOX11 syndrome.

Outcomes and results:

Most were found to have borderline (33 %) or mild (39 %) impairment in adaptive behaviour, with more difficulties in communication and daily living than socialisation in the cohort overall. Most (90 %) were found to exhibit clinically relevant levels of autistic traits, with 62 % scoring in the "severe" range, though social motivation was observed to be a relative strength in the cohort overall.

Conclusions and implications:

This study presents the first standardised evaluation of adaptive behaviour and autistic traits of individuals with SOX11 syndrome. This will improve clinicians, educators and parents' understanding of SOX11 syndrome.