The Developmental Consequences of Rare Chromosome Disorders:
Two Case Reports

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**Aim:** As molecular and cytogenetic testing becomes increasingly sophisticated, more individuals are being diagnosed with rare chromosome disorders. Yet despite a burgeoning knowledge about biomedical aspects, little is known about implications for psychosocial development. The scant literature gives a general impression of deficits and adverse developmental outcomes.

**Method:** Developmental data were obtained from two 16 year olds diagnosed with a rare chromosome disorder – a girl with 8p23.1 and a boy with 16q11.2q12.1. Measures of intellectual ability, academic achievement, and other aspects of functioning were administered at multiple time points from early childhood to adolescence.

**Results:** Both adolescents experienced initial delays in motor and language development. Although the girl's intelligence is assessed as being in the average range, she experiences difficulties with motor planning, spelling and writing. The boy has been diagnosed with a mild intellectual disability and demonstrates mild autistic features.

**Conclusions:** The two case descriptions are in marked contrast to the published literature about these two chromosome anomalies. Both adolescents are developing much more positively than would be expected on the basis of the grim predictions of their paediatricians and the negative reports in the literature. It is concluded that, for most rare chromosome disorders, the range of possible developmental outcomes is currently unknown.