Variable phenotype in 16p duplication within a family

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Background.  
More individuals are now being identified with very rare genetic syndromes. We present a family with an inherited duplication of 16p11.2 to 16q12.1 in ring formation. Three of the four children, (aged 15 months to 10 years), mother, uncle, and grandmother are affected. Our aim was to provide preliminary evidence of possible phenotypic patterns of learning and behaviour associated with this chromosome anomaly.

Method.  
Psychometric assessments were undertaken with all four children. The mother and uncle also agreed to participate in the study. Measures of development (Bayley or Mullen), intellectual ability (WISC-IV or WAIS-III), academic achievement (WIAT-II), adaptive behaviour (Vineland), and other relevant aspects of functioning (e.g., Children’s Memory Scale) were administered.

Results.  
The first-born child is the only one who is unaffected. Her intellectual ability was assessed as being within the superior range. The second child experienced early difficulties with speech and motor skills. Although his intelligence is average, he has learning difficulties and significant auditory memory problems. The third child’s speech and motor milestones were markedly delayed. He has a complex medical history that includes a vitamin B12 deficiency. On the Mullen Scales at age 4 his scores ranged from average to very low. The development of the youngest child (aged 15 months), who also had a B12 deficiency but was treated early, was assessed as being within typical limits.

Conclusions.  
There is considerable developmental variability among the three children with this inherited 16p duplication. We discuss the intriguing similarities and differences, considering common features that may reflect phenotypic patterns and speculating about possible explanations for the variable presentations.

Keywords: 16p duplication; behavioural phenotype; rare chromosome disorders