Screening for fragile X syndrome

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Record Status

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Authors' objectives

Aims to provide the information needed to decide whether to use DNA testing to screen for the fragile X syndrome, a common cause of learning disability.

Authors' conclusions

Limited paediatric screening for fragile X syndrome and some cascade screening in affected families is currently being carried out at many UK centres. This is of clinical value and should continue. However, more research will be needed before any active screening programmes should be considered for implementation in the NHS.

Studies should be carried out to assess the current practice of paediatric screening when there is developmental delay.

There should be a national audit of the current practice of screening in affected families.

Research should be commissioned into the psychosocial implications of being identified as having a PM.

The feasibility of routine antenatal screening should be assessed. A central register for all diagnoses should be established, based mainly on reports from DNA laboratories.

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