



Title	Screening for Fragile X Syndrome: A Literature Review and Modelling Study
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Aim

To compare the effectiveness, estimate the associated costs, and summarize available evidence about the feasibility and acceptability of different screening strategies in England and Wales. To establish a model for estimating effectiveness and costs of these different strategies.

Conclusions and results

Simulation results by the FXS Model showed that, over the first 10 years, 4% of premutation (PM) females and 70% of full mutation (FM) females could be detected by active cascade screening (versus 10% and 58%, respectively, by prenatal screening). The maximal detection rate for FM carriers by active cascade screening is higher than that by prenatal screening (91% versus 71%). However, the maximal rate of detection of female PM carriers by active cascade screening (6%) is much lower than that by prenatal screening (60%). During the first 10 years of simulation, the estimated direct cost per year to the NHS in England and Wales is £0.7–0.2 million by active cascade screening and £14.5–9.1 million by a program of prenatal screening. The incremental cost per extra carrier detected (using current practice as the reference standard) is, on average, only £165 by active cascade screening versus £7543 by prenatal screening. The incremental cost per FXS birth avoided is, on average, £8494 by active cascade screening versus £284 779 by prenatal screening.

Recommendations

The empirical evidence suggested that both prenatal screening and cascade screening are feasible and acceptable. They both can reduce births of FXS children and save cost in the long term. Population-based prenatal screening is more efficacious and has a greater impact on the population, but also costs more than active cascade screening. The active cascade screening of affected families is more efficient, cheaper, but less effective than population-based prenatal screening.

Methods

Assessed the published literature. Efforts focused on developing a model that could be used to synthesize data from various sources, estimate the cost effectiveness of different strategies, and conduct sensitivity analyses according to different assumptions.

Further research/reviews required

It is suggested that both strategies be evaluated in large-scale trials, which might also help to determine whether and how the different strategies could be simultaneously or sequentially combined.