Implementation of a national prenatal exome sequencing service in England: cost-effectiveness analysis

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Abstract

Objective To evaluate costs and cost-effectiveness to the healthcare system, and costs to families, of a national prenatal exome sequencing (pES) service additional to standard testing, compared to standard testing alone. **Design** A cost-effectiveness analysis combining costs, outcomes, parent and professional interview and professional survey data. **Setting** The English National Health Service (NHS) Genomic Medicine Service. **Sample** 413 cases referred for pES testing from 01/10/2021 to 30/06/2022. **Methods** We costed the incremental resource required to deliver the pES clinical pathway, synthesising this with unit costs and outcomes data on additional cases diagnosed to analyse cost-effectiveness. We estimated the annual NHS budget requirement based on case numbers. We determined parental costs from interviews. **Main Outcome Measures** Incremental costs of pES to the NHS and families, incremental cost per additional diagnosis, NHS budget impact. **Results** Of 413 referred cases, 241 were tested, at a cost of £2,331 (95% credibility interval £1,894-£2,856) per referred case, or £3,592 (£2,959-£4,250) per case that proceeded with testing. The incremental cost per diagnosis (yield 35.3%) was £11,326 (£8,582-£15,361). At current demand levels pES costs the NHS approximately £1.7m annually. Family costs could not be separated from other pregnancy related appointments but were not considered burdensome as most appointments were concurrent or remote. **Conclusions** pES is more expensive than predecessor prenatal genetic testing technologies, has a higher diagnostic yield and informs pregnancy management and decision making. Further research into potential savings from the foregone diagnostic odyssey resulting from pES may be informative.

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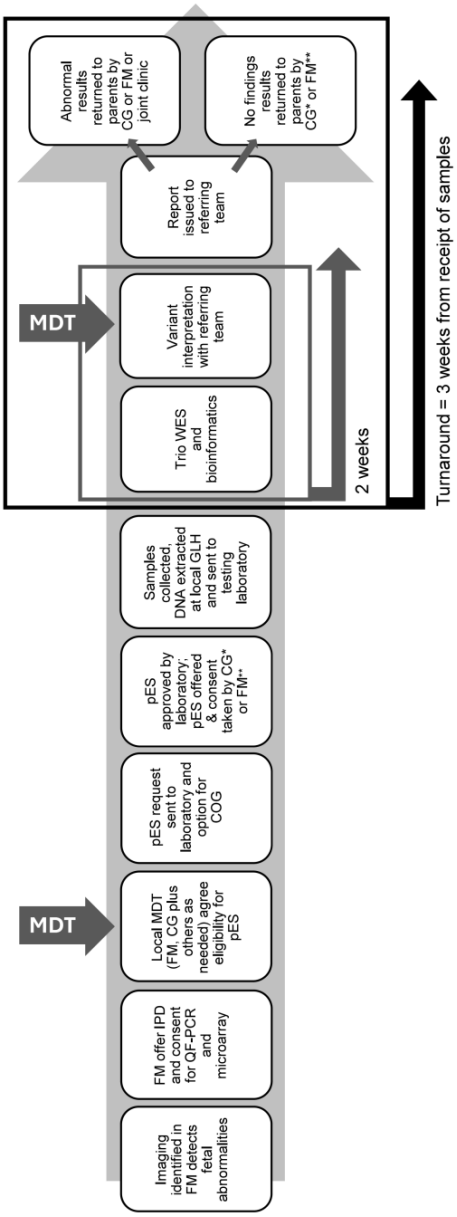

Figure 1. Overview of the pES pathway. Local pathways can vary in which staff groups are involved in taking consent and return of results.
MDT = Multi-disciplinary team, FM = Fetal medicine, IPD = Invasive prenatal diagnosis, QF-PCR = quantitative fluorescent-polymerase chain reaction, CG = Clinical genetics, COG = Clinical oversight group, GLH = Genomic laboratory hub, WES = Whole exome sequencing; * = may include genetic counsellors; ** = may include midwives

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