

Prenatal whole exome sequencing: the views of clinicians, scientists, genetic counsellors and patient representatives

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OBJECTIVE: Focus groups were conducted with individuals involved in prenatal diagnosis to determine their opinions relating to whole exome sequencing in fetuses with structural anomalies.

METHOD: Five representatives of patient groups/charities (PRGs) and eight clinical professionals (CPs) participated. Three focus groups occurred (the two groups separately and then combined). Framework analysis was performed to elicit themes. A thematic coding frame was identified based on emerging themes.

RESULTS: Seven main themes (consent, analysis, interpretation/reinterpretation of results, prenatal issues, uncertainty, incidental findings and information access) with subthemes emerged. The main themes were raised by both groups, apart from 'analysis', which was raised by CPs only. Some subthemes were raised by PRGs and CPs (with different perspectives). Others were raised either by PRGs or CPs, showing differences in patient/clinician agendas.

CONCLUSIONS: Prenatal consent for whole exome sequencing is not a 'perfect' process, but consent takers should be fully educated regarding the test. PRGs highlighted issues involving access to results, feeling that women want to know all information. PRGs also felt that patients want reinterpretation of results over time, whilst CPs felt that interpretation should be performed at the point of testing only

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