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Short report

Will genetic testing for predisposition for disease result in fatalism? A qualitative study of parents responses to neonatal screening for familial hypercholesterolaemia

Victoria Senior <sup>a</sup> ... Timothy J Peters <sup>b</sup>

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Abstract

**Objective:** to describe parents' perceptions of familial hypercholesterolaemia (FH), an inherited predisposition to heart disease, following population-based neonatal screening.

**Design:** a qualitative analysis of semi-structured interviews with the parents of 24 children who had received a positive screening test result informing them that their child was at-risk for having FH.

**Results:** responses to screening seemed to vary according to perceptions of the

underlying cause of the positive screening test result. When parents perceived the test as detecting raised cholesterol the condition was perceived as familiar, dietary in origin, controllable and less threatening. When the test was seen as detecting a genetic problem, the condition was perceived as uncontrollable and, hence, more threatening.

Conclusion: these pilot data raise questions about the extent to which assessing disease risks by DNA analysis may result in a sense of fatalism, adversely affecting motivation to change behaviour and to reduce risks.



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## Keywords

Genetic testing; Neonatal screening; Illness perceptions

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