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Better diagnosis for people with genetically raised cholesterol

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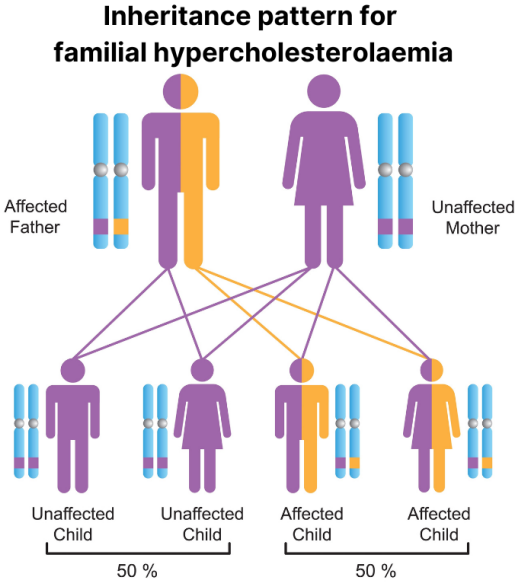

Using family networks - “cascade testing” - is a potentially important way to diagnose people with genetic conditions. However, this approach raises questions around how best to engage and test families to improve diagnosis and treatment rates. We explored these issues in the context of familial hypercholesterolaemia. This is an inherited condition that causes raised cholesterol from birth and increases the risk of heart disease if left untreated.

We developed cost-effectiveness models using routine data from primary care and existing cascade testing services, to assess the costs and benefits of different ways of designing cascade testing services.

We found that the most cost-effective approach to cascade testing involved health care professionals directly contacting relatives of the person with familial hypercholesterolaemia and that more distant relatives should still be contacted even if close relatives don’t engage with the cascade testing process. We also found that similar health outcomes and costs could be achieved by using a combination of cholesterol testing and confirmatory genetic testing, or by using genetic testing exclusively. These findings suggest that health services can tailor the testing process to reflect local service arrangements and constraints without compromising on the

delivery of effective and cost-effective care.

Our results can help health care decision makers to design effective and cost-effective approaches to cascade testing. In turn this has the potential to greatly improve the diagnosis of familial hypercholesterolaemia in the UK.

Another important question is how and when to treat children and young people diagnosed with familial hypercholesterolaemia. We have started a new project to address this question.

Read the full paper in Health Technology Assessment and related work in Atherosclerosis.

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