MedicalXpress

Creating a better understanding of recessive genetic disorders

September 7 2011

Researchers from the University of York are investigating why so few fathers are tested for sickle cell and thalassaemia during ante-natal care when their partner is a known carrier.

The disorders are the UK's most common recessive genetic conditions – both parents must carry the trait before it can be passed on to their child – with at least 14,000 people affected.

Both conditions affect the red blood cells containing the protein haemoglobin which carries oxygen from the lungs to all parts of the body. Thalassaemia can lead to severe anaemia requiring lifelong treatment. In sickle cell disorders, severe pain and damage to organs such as the liver, kidney, lungs, heart and spleen are common.

Researchers led by Professor Karl Atkin from the University's Department of Health Sciences, have been awarded a £233,000 Research for Patient Benefit (RfPB) grant to explore why - despite the introduction of a Sickle Cell and Thalassaemia Screening Programme in 2001 by the Department of Health - so few fathers are tested.

Professor Atkin said: "If a mother is identified as a carrier, her partner should ideally be offered screening, enabling the couple to make an informed choice on the future of the pregnancy. However, recent evidence suggests few fathers are tested even when their partner is a known carrier. Little is known about why.

“This research, involving focus-group discussions with prospective mothers and fathers, interviews with fathers who are likely to have been involved in their partner’s ante-natal process, and discussions with practitioners and commissioners, will explore the reasons for the poor uptake.”

Researchers will be working closely with the NHS Screening Programme, healthcare professionals, voluntary organisations and local communities throughout the “Involving fathers in ante-natal screening for sickle cell disorders: Improving informed decision” project. The research findings will be used to develop more sensitive policy and practice and better engagement with fathers during the ante-natal screening process.

There are an estimated 240,000 sickle cell and 214,000 thalassaemia trait carriers in the UK. Those most at risk of sickle cell disorders are of African-Caribbean or African origin, while thalassaemia is more commonly found among Cypriots, South Asian and Chinese populations. However, Professor Atkin says ethnicity is becoming less reliable as a predictor of who carries a trait.

“National screening programmes have identified far more ‘white’ carriers than expected and the increasing proportion of the population who claim ‘mixed heritage’ further complicates the situation,” he said.

Where both the mother and father are carriers, there is a one in four chance that their child will be born with the disorder and a fifty per cent chance that their child will be a carrier.

York’s Department of Health Sciences has also received a further £230,000 from the Economic and Social Research Council (ESRC) to investigate the social consequences of being a trait carrier.

The project, 'Living with sickle cell or beta thalassaemia trait', also led by Professor Atkin, will involve talking to carriers of different ages about how the trait affects their day-to-day living, family relationships and social networks, and will concentrate on reproductive choice and family formation.

Professor Atkin said: "There is very little research available on the social consequences of being a 'healthy' trait carrier for a recessive disorder, particularly at different stages of the life cycle. Our findings will contribute to theoretical debates as well as policy and practice, and will be used to make suggestions about developing potential support, enabling people to make informed decisions about their health."

Provided by University of York

Citation: Creating a better understanding of recessive genetic disorders (2011, September 7) retrieved 10 April 2024 from https://medicalxpress.com/news/2011-09-recessive-genetic-disorders.html