Tay Sachs Disease carrier screening in the Ashkenazi Jewish population

Tay Sachs disease (TSD) is a genetic form of degenerative neurological disease caused by mutations in the hexosaminidase A (*HEXA*) gene. The most common form of the disease is lethal in infancy or early childhood, and there is no effective treatment. TSD is inherited in an autosomal recessive manner, meaning that individuals with one copy of a mutation in the *HEXA* gene are 'carriers' - healthy, but at risk of passing on the mutation to their children; individuals who inherit two copies of *HEXA* gene mutations will develop the disease. The chance of two healthy carriers of mutations in the *HEXA* gene having a child affected by the disease is one in four.

The carrier frequency for TSD is substantially increased in Jewish populations of Ashkenazi origin (originating from Central or Eastern Europe, around 95% of British Jews) compared with the general population. Carrier screening for TSD in the Ashkenazi Jewish population has been available in the UK since the 1980s, and funded by the National Health Service (NHS) since 1999. Biochemical or molecular (DNA) carrier testing allows individuals identified as carriers to reduce their risk of having a child affected by TSD, and there is widespread acceptance of TSD screening by Jewish communities in the UK, as well as internationally.

In a joint project commissioned by the UK Newborn Screening Programme Centre, a team from Guy's and St Thomas' Clinical Genetics Service in London and the PHG Foundation in Cambridge, working with an expert Advisory Group, undertook a needs assessment and review of current TSD screening services in the UK. The final report from this project, *Tay Sachs Disease carrier screening in the Ashkenazi Jewish population*, sets out their findings. These include:

- **Carrier screening can be provided within the community and in the antenatal setting but is generally not offered in a systematic way**
- **Carrier screening is accessed via clinical genetics services, two walk-in clinics in London, and outreach community screening provided by Jewish voluntary organisations.**
- **Carrier testing is performed at NHS laboratories in London (Guy's) and Manchester (Willink).**
- **A survey of antenatal screening provision showed that in the 114/155 hospital trusts that responded, screening was not routinely offered even in areas with large Jewish populations; there was general confusion about who should be offered testing, and a common assumption that Jewish women had already been tested.**
- **A patient survey showed that most were aware of their Ashkenazi Jewish ethnic origins, 97% having four Jewish grandparents, but had heard about screening primarily via family, friends or the internet rather than from midwives or general practitioners. One third of respondents were already pregnant.**

The Report concludes with a set of recommendations that could be used to strengthen the NHS carrier testing offered to the Ashkenazi Jewish population.

PHG Foundation Programme Director and report co-author Dr Hilary Burton commented:

"The National Screening Committee and providers of genetics services should be in no doubt that Tay Sachs disease carrier screening is an extremely important health service that is both needed and wanted by the Jewish community. Throughout the world, where there are extensive Jewish communities, carrier screening programmes are in place and have been extremely effective in reducing the birth prevalence of this devastating condition.

It is only because the Jewish community in the UK is relatively small in size and has been proactive in raising awareness and providing screening through the voluntary and private sector that the number of affected Jewish children in recent years has been quite few. NHS maternity services, primary care, genetics services and their commissioners, especially in areas with large Jewish populations, all have a role to play in bringing this continuing risk to the attention of Jewish individuals and couples and providing access to high quality carrier testing services. This Report provides all the necessary background for action."

Dr Ian Ellis, Consultant Clinical Geneticist at the Alder Hey Children's Hospital in Liverpool and member of the project expert advisory group commented:

"People have to realise that Tay-Sachs has not gone away, but only through constant vigilance and screening can families be made aware of the potential risk and their options. This report documents the previously haphazard arrangements for Tay Sachs screening in Britain. The report is comprehensively written and provides an opportunity for building Tay-Sachs screening into current NHS screening plans. There needs to be a coordinated and comprehensive plan so that those in at-risk UK Jewish community who wish to access screening, can do so easily and reliably.

[The report] should remove the fear, fallacies and hopefully the funding dilemmas to provide a comprehensive, quality NHS funded partnership between trained genetic counsellors, quality assured genetic screening laboratories and Jewish community agencies for education."

The report is available for free download from the PHG Foundation website, **www.phgfoundation.org**

- Ends -

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Notes to Editors

ADDITIONAL INFORMATION, 21st May 2009

- Dr Anne Mackie, Programme Director for the UK National Screening Committee, has stated: *"The UK NSC believes that antenatal Tay Sachs testing should be available for individuals from high risk populations - including the Ashkenazi Jewish population - who seek it. We are asking NHS commissioners to assess the need for this service in their area and provide a service to those who have a reason to seek it."*
- Website: http://www.screening.nhs.uk/taysachs; contact NSC Press Office tel: 020 7400 7390 or email: nscpressoffice@hanovercomms.com

PHG Foundation

- The PHG Foundation is an independent, not-for-profit, multidisciplinary health policy research and development organisation based in Cambridge, UK, but working internationally.
- We focus on 'bridging the gap' in clinical translation to move new genomic and biomedical knowledge and applications into health service practice and better health for people and populations.
- We are the leading UK centre for public health genomics, and have considerable experience in policy research and development, as well as evidence-based evaluation of clinical tools and health services.
- Our expertise in areas including the scientific, clinical, public health, ethical, legal, social and regulatory aspects of genomics and biomedicine.

The PHG Foundation is the working name of the Foundation for Genomics and Population Health, a charitable company registered in England and Wales, charity no. 1118664 / company no. 5823194.

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Guy's and St Thomas'

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- Website: www.guysandstthomas.nhs.uk.
- Contact: Malcolm Bennie, Guy's and St Thomas' NHS Foundation Trust tel: 020 7188 1553 or email: malcolm.bennie@gstt.nhs.uk. Out of hours, please call our pager bureau on 08700 555500, ask for pager number 847704 and give the pager operator your message.