Enhanced Genetic Services Project

Evaluation Report

Executive Summary
The former Heart of Birmingham Primary Care Trust (HoBtPCT) had identified in November 2008 that autosomal recessive (AR) conditions were contributing to the excess infant mortality rate in Birmingham, and in particular of that experienced by the population served by the PCT. The Enhanced Genetic Services Project (EGSP) was established by HoBtPCT to address excess infant mortality and childhood morbidity in Birmingham linked to these conditions.

Compared to other ethnic groups, the Pakistani community had been identified as being at particular high risk of AR conditions. These conditions occur more frequently in communities where consanguineous partnerships are common. EGSP focused on identifying those families with a history of AR conditions, and who would benefit from individual genetic risk information, to enable couples to make informed reproductive choices. The Pakistani community was identified as the main focus of EGSP but other communities with high rates of AR conditions were also included in the Project. These activities were complemented by a primary care strand and an education strand.

The West Midlands Perinatal Institute provided data for the evaluation on infant mortality and stillbirth rates for the population of Birmingham, which were broken down by ethnic group and by cause of death. This detailed information was not routinely available prior to the initiation of EGSP. Their analyses suggested that although infant mortality rates had decreased, the population of Birmingham continued to experience elevated rates of infant deaths and stillbirths compared to the England and Wales average and that mothers of Pakistani origin experienced higher rates compared to the White European reference group. Babies from this community were at higher risk of being affected by a congenital anomaly compared to the reference group and were at particularly high risk of being affected by an AR condition.
EGSP components

EGSP was set up comprising three strands and an evaluation.

**Strand 1 – Primary care**
Strand one was located within primary care services and involved establishing opportunistic screening for haemoglobinopathies within pilot GP practices and using this as a vehicle to raise genetic literacy amongst health care staff and the patient population. Two practices participated in EGSP. Testing was offered to the practice population aged 17 and over.

**Strand 2 – Clinical genetics services**
Strand two consisted of an extension of the services offered by the West Midlands Clinical Genetics Services, and included the systematic identification of families appropriate for EGSP genetic services. This strand involved two phases: firstly, a review of all existing families from Birmingham known to the clinical genetics department (to establish the range and number of families with known AR conditions), followed secondly by case finding, which was then to be extended through other medical specialties. This second phase included the development of increased capacity of the local molecular genetics laboratory to provide genetic testing. There was the intention to provide outreach genetic counsellor-led clinics in local GP practices.

**Strand 3 - Education**
This comprised educational initiatives in the community and with health care professionals. Educational materials were developed to support the educational activities, and as a tool for individuals to approach and provide information for their extended families.

**Strand 4 - Evaluation**
This was an integral part of EGSP and included ongoing evaluation support for the project by an externally commissioned organisation. The evaluation was to provide iterative feedback to inform the ongoing running of the project as well as the final evaluation report. The PHG Foundation was commissioned to provide this support for EGSP and established the Evaluation Working Group.

**Project objectives**
**Objective 1:** Improved access to molecular testing for autosomal recessive disorders.

**Objective 2:** Improved access to genetic counselling through increased capacity within the clinical genetics service of genetic counsellors with appropriate minority languages.
Objective 3: Improved genetic literacy in families affected by a genetic disorder identified by the initiative.

Objective 4: Improved genetic literacy and understanding in the extended families.

Objective 5: Improved genetic literacy in HoBtPCT’s ethnic minority communities about genetic disorders and their transmission, particularly in consanguineous families.

Objective 6: Improved genetic knowledge and competence among health professionals in HoBtPCT.

Objective 7: Carrier testing within extended family to identify high risk couples, offering genetic counselling including possibility of prenatal diagnosis and postnatal treatments.

Objective 8: Identify causative mutations in genes known to cause autosomal recessive conditions in affected families.

Objective 9: Offer carrier testing of causative mutations to the extended family.
Conclusions arising from strand activities

Primary care

- The two practices identified three patients affected by thalassaemia, 99 thalassaemia carriers, eight patients affected by sickle cell disease and 43 sickle cell disease carriers.

- Careful consideration needs to be given to the information patients needed in order to understand the impact of being affected by or being a carrier of a genetic disorder on themselves and the implications for other family members. Without the provision of this information the benefits of such a screening programme will not be fully realised.

- It is necessary to ensure systems are in place to support all aspects of a new patient care pathway. In particular, fail safe mechanisms need to be established prior to initiating new developments, including pilot schemes.

Clinical strand

- EGSP achieved the delivery of services set out in the pathway documents. Two hundred and fifty sets of patient notes were reviewed, which led to the EGSP re-contacting 80 individuals and offering them genetic counselling. Of these, 55 individuals actually received genetic counselling. Thirty six individuals were offered genetic testing, of whom 26 accepted the offer.

- One hundred and eighty-eight families were reviewed and potentially over 700 family members were identified as ‘at risk’, with the potential to benefit from genetic counselling and testing. Almost 400 family members were discussed with the proband or the proband’s parents resulting in the proband making known contact with 37 family members. By the end of the Project, 56 family members had been offered and had accepted genetic testing, and 169 families had been identified with eligible conditions for EGSP services.

- In practice, cascade testing was limited to immediate family members. One hundred and twenty-one children, such as siblings under the age of 16, were identified who at a later stage may decide to take up testing. It is unclear how far cascade testing is successful in routine clinical genetics practice, or whether, as in the case of EGSP, cascade testing is usually limited to the immediate family.

- EGSP services were well received by patients. They enhanced genetic understanding and facilitated sharing of genetic information with other ‘at risk’ family members.

- Thirty-one genetic tests, including five diagnostic and 26 carrier tests were undertaken for individuals meeting EGSP criteria, using EGSP developed tests. In addition 185 diagnostic tests and 75 carrier EGSP developed tests were undertaken for individuals living in the West Midlands but not meeting EGSP criteria.
O Five EGSP patients received a molecular diagnosis using EGSP developed tests and 17 had carrier status confirmed. Using non EGSP developed tests, a further five molecular diagnoses were made and 29 positive carrier status were confirmed for EGSP patients.

O A total of 111 molecular diagnoses were made for non-EGSP patients using EGSP developed tests and 92 positive carrier status confirmed for this group of patients.

O New genetic tests have been developed; they have generated sufficient income from outside the West Midlands for the service to be sustained beyond the EGSP funding.

Education strand

O For in-depth community education on genetic risk that goes beyond awareness raising and enables specific concerns to be addressed, education is usually best delivered on a one-to-one basis. Similar programmes should incorporate both group work and the ability to work on a one-to-one basis. This highlights the importance of an interlinked clinical and education strand, so that individuals who require further information and support, and potentially genetic testing, have access to this delivered by a genetic counselling service.

O Community and professional education is a process that requires sustained efforts for educational initiatives to be delivered. Repeated contacts are often necessary following an initial approach.

O Education sessions in the community had a snowball effect when further sessions were planned on subsequent weeks. Women would return with further questions and with others that they felt would benefit.

O Consanguinity can be an emotive issue but if sensitively dealt with in a manner that is nuanced and conveys an accurate understanding of the risks, it need not be controversial. In the experience of EGSP, there were no negative responses from the community to the project. Initially some level of suspicion was often encountered, but if dealt with sensitively did not persist. Members of the community valued the services provided by EGSP, including a fuller understanding of risks.

O GP practices need to have the expertise to advise people on risks associated with consanguinity and AR conditions. Members of the community turn to their GP for such information. GPs often do not see genetics training as a priority and few had made referrals to the clinical genetics service.

O It is important to identify the approach to education that will result in most engagement and achieve best outcomes. The initial focus on genetics was less effective than focusing on high rates of infant mortality and morbidity in the locality. Once the initial interest had been developed, the audiences were more likely to engage with other aspects of education such as genetics, AR inheritance and how consanguinity can increase risk in certain families.
Recommendations

1. Autosomal recessive conditions are an important contributor to the excess stillbirths and infant mortality rates in the Birmingham Pakistani and Bangladeshi communities. This contributing factor should be considered and addressed as part of public health efforts to address health inequalities in these populations.

2. Further work is required to establish the evidence for the appropriate model of service in primary care to provide care to communities at high risk of stillbirths and infant mortality deaths due to autosomal recessive conditions. This should include ensuring adequate systems are in place to support any new patient care pathways and to provide fail safe mechanisms.

3. Long-term enhanced public health surveillance of excess stillbirths and infant mortality rates in the West Midlands based on the systems and data generated from this Project should be established to provide the evidence base for identifying and addressing the contributory causes.

4. The learning and experience of the project should be embedded into the West Midlands Clinical Genetics Service to ensure enhanced cascade testing is available for families with identified risk of autosomal recessive diseases.

5. Genetic counsellor outreach services into general practice should be considered for a pilot, particularly in those practices with the highest population risk of autosomal recessive conditions.

6. The West Midlands Regional Genetics Laboratory should continue to develop new genetic tests, particularly for autosomal recessive conditions, building on the capacity and experience generated from EGSP and the opportunities provided by next generation sequencing technology.

7. Future health development work should build on the achievements and materials developed by the education strand including the project website. Expertise developed by the EGSP professional and community education strands should be embedded into routine professional education and community education initiatives.

8. The Health Visitor service should nominate a lead health visitor for clinical genetics and this lead role should be supported by the West Midlands Clinical Genetics Service.

9. The development of patient support such as ‘Patient Champions’ should be considered as part of future programmes to improve both community and professional awareness and to support cascade testing.

10. The burden of morbidity associated with AR conditions needs to be formally investigated and described as a matter of urgency. This should include quantifying the range of health and social care consequences and their associated costs.