EGSP
Enhanced Genetics Services Project

An overview of the Enhanced Genetics Services Project in Birmingham

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EGSP
Enhanced Genetics Services Project

What will be covered in the workshop?

Setting the scene

- ❑ **General overview of the service**
- ❑ **Background**
- ❑ **Aims of EGSP**
- ❑ **Benefits of the service**
- ❑ **Team**
- ❑ **Referral information**
- ❑ **Outcomes at present**

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…..and most importantly your input too!

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Background

- Genetic disorders are known to contribute significantly towards the high perinatal mortality rates in Birmingham and are particularly common in the children of couples who have married within the family.
- Raising awareness of genetics and the genetics services in the community is necessary in order to enable families to take full advantage of new developments in diagnostic tests and potential treatments
- Most importantly to make informed choices.

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Background

- Former Heart of Birmingham PCT teaching Trust had recognised that Autosomal Recessive (AR) conditions were contributing to the excess infant mortality rate in Birmingham, particularly for the local population.
- Reducing infant mortality and perinatal mortality had been a key health target.
- The trust through a mutual understanding allocated funds and delegated the responsibility to WMRGS to raise awareness and promote genetic services to the local population.
- As a result of this a 3 year project was funded 'Enhanced Genetics Services Project'.
- EGSP was established to address excess infant mortality and childhood morbidity in Birmingham linked to AR disorders.

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Background

- **Autosomal Recessive (AR) conditions occurred more frequently in communities where consanguineous partnerships are common.**

The continuity popularity of consanguineous unions in many present day and urban populations is apparent from the detailed information in the Global Consanguinity Database (www.consang.net).

To put this into context:

***Intra-familial unions between couples related as second cousins or closer are favoured in regions such as North and Sub-Saharan Africa, the Middle East, and Central and South Asia and among many emigrant communities from the region reside in Europe and America* (Bittles, 2012 pg 3).**

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Aims of the project

1. Raise awareness of the genetic services in Birmingham.
2. To improve the community and health care professionals understanding of inheritance and EGSP project.
3. Encourage and increase referrals to the service.
4. Developing tests for conditions.
5. Communication with stakeholders
6. Contribution towards a possible reduction in infant mortality.

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- The most recent data from BPH shows that the infant mortality rates in Birmingham was 7.3 per 1000 births between 2008-2010 compared to 4.6 per 1000 births over the same period for England.
- During the same period, the Perinatal mortality rate for Birmingham was 11.0 per 1000 births compared to 7.5 per 1000 for England, and the stillbirth rates 6.9 per 1000 births in Birmingham, compared to 5.1 per 1000 births in England.

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What are the benefits of the service?

1. In the long term, a reduction in mortality and morbidity due to genetic disorders in the communities that are most affected.
2. Greater understanding of genetic disorders and options available throughout affected families.
3. Improvement in the community level of genetic literacy (GP and Educational Strand).
4. Increase in understanding of genetics of health professionals (likely to impact outside the remit of the current study).
5. Cost saving coupled with better provision of services by batching genetic testing.

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Where is the EGSP based?

- West Midlands Regional Genetics Services in Birmingham Women's Hospital.
- Has outpatient department in Norton Court.
- Outpatient clinics within Birmingham areas also other parts of the West Midland region.
- Genetic counsellors and Genetic Consultants work in and around Outpatient clinics within Birmingham areas also other parts of the West Midland region.

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How does the project work: EGSP is inclusive of three strands

Based at the Clinical Genetics Unit at BWH

Stand 1-The Clinical Strand

- We have reviewed and re-contacted families already known to paediatricians and the clinical genetics service to update them with new relevant information and offer new services and treatments where appropriate.
- This has been supported by the development of new molecular diagnostic services within the West Midlands Regional Genetics Laboratory.
- The molecular testing services has been developed by laboratory staff and supported by genetic counsellors and lead consultant geneticist (Dr Williams).
- It was intended that an outreach service would be provided by Genetic Counsellors in local GP practices, however this was not viable.

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Disease	Genes	UKGTN gene dossier
Niemann Pick disease type C	NPC1	Y
	NPC2	Y
Gaucher disease	GBA	N
Progressive familial intrahepatic cholestasis	ATP8B1	Y
	ABCB11	Y
	ABCB4	Y
Citrin deficiency	SLC25A13	Y
Pompe disease	GAA	N
Metachromatic leukodystrophy	ARSA	N
Saposin	PSAP	N
Krabbe disease	GALC	N
Sanfilippo syndrome	SGSH	N
	HGNSAT	N
	NAGLU	N
	GNS	N
Fowler syndrome	FLVCR2	Y
Propionic acidaemia	PCCA	Y
	PCCB	Y
Glutaric acidaemia	GCDH	N
Morquio A syndrome	GALNS	N
I-cell disease	GNPTAB	Y
Argininosuccinic aciduria	ASL	N
ASPM microcephaly	ASPM	Y
Isovaleric aciduria	IVD	N
Citrullinaemia type I	ASS1	N
Succinic semialdehyde dehydrogenase deficiency	ALDH5A1	N
Niemann Pick disease type A/B	SMPD1	N
Homocystinuria	CBS	N
	MTHFR	N
Trichohepatoenteric syndrome	TTC37	Y

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Strand 2-The education Strand (Community & Clinical)

- **This strand includes educational initiatives both in the community and with health care professionals.**
- **In the community the aim is to improve genetic literacy and with health professionals the aim is to develop educational competences to enable families to be supported with information, advice and signposting to appropriate local specialised services.**
- **The project also provides holistic and culturally sensitive information when working closely with the community and extended family members of affected individuals to improve their understanding of hereditary conditions and help them access new services/developments.**
- **Educational leaflets have been developed to be used in the community strand (please see resources on table).** www.talkinggenetics.co.uk

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Community strand

When asked how confident the attendees were to share genetic information with their family before reading Leaflet 3. The table below shows that confidence levels in discussing genetic information with their families dramatically increased.

	Very Confident	Confident	Confident but I need more information	Not confident
Confidence in sharing genetic information with your family before reading Leaflet 3	1	3	10	49

	Very Confident	Confident	More confident but I need more information	Not confident
Confidence in sharing genetic information with your family after reading Leaflet 3	22	31	10	0

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Attendees were also asked where they would go to get further information on genetics and the answers were varied. The results demonstrated that members of the community are most likely to approach their GP or access the internet for further information. This clearly reinforces the role of providing supportive education to health care professionals such as GP's as being detrimental in promoting genetic services.

Comments such as the service is easy to find/access and user friendly website. (Please note, some attendees wrote more than one answer);

GP:	12
NHS:	2
Internet / Website	29
EGSP	8
Hospital	1
Support Group	6
Library	2
Hospital	1
Other / Question not answered	4

Take family member to see GP	7
Talk with them and show them the leaflets	27
Talk with them and show them the website	16
Discussing / Talking with them	26
Explain in my language	4
Speak to elder to speak to the rest of the family	1
Question not answered	3

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Cont

Attendees were asked how they would personally share genetic information with their families, the responses were as follows (Please note that some attendee's provided more than one answer);

Take family member to see GP	7
Talk with them and show them the leaflets	27
Talk with them and show them the website	16
Discussing / Talking with them	26
Explain in my language	4
Speak to elder to speak to the rest of the family	1
Question not answered	3

When asked how important the information on leaflet 3 was to consanguineous families 63/63 attendees answered that it was 'very important'

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Do you feel it is important to raise awareness of the risks of genetic disorders within families?				Has it been a useful way to raise awareness?			
Yes	12	No	0	Yes	12	No	0

Comments
We want them to have healthy children'
"You can't change arranged marriages to cousins but it's worth talking to them about the risks to their children'
'I have relatives who are married to their cousins who have many children with disabilities'
'I am seeing a Somali lady at the moment who has had lots of miscarriages and is married to her first cousin, she is pregnant'

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Do you feel it is important to raise awareness of the risks of genetic disorders within families?	Has it been a useful way to raise awareness?

Comments :

It is important to raise awareness as many people do not know about this'

'You don't get to hear about this much'

of course it is important but you can't change some peoples way of thinking'

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Strand 2-The education Strand (Clinical Education)

- **The project provides the necessary training for non-genetic health care professionals to recognise families that may be at risk of inherited disorders and know how to refer appropriately.**
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Strand 3- The GP Strand

Three GP Practices in the former Heart of Birmingham Primary Care Trust have participated in this project and have offered a screening programme for inherited blood disorders. They are raising awareness of other genetic disorders and are identifying families appropriate for specialist referral.

This has involved in establishing opportunistic screening for Thalassemia and raised genetic literacy amongst health care staff and patients. Testing was offered to the practice population aged 16 and over.

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How will the EGSP be evaluated?

This has been a core role within the project. An external organization called PHG foundation are providing ongoing evaluation of the project. The aim of the evaluation is to provide feedback to inform the ongoing project as well a final evaluation.

(Foundation for Genomics and Population Health) based in Cambridge.

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Families with genetic conditions

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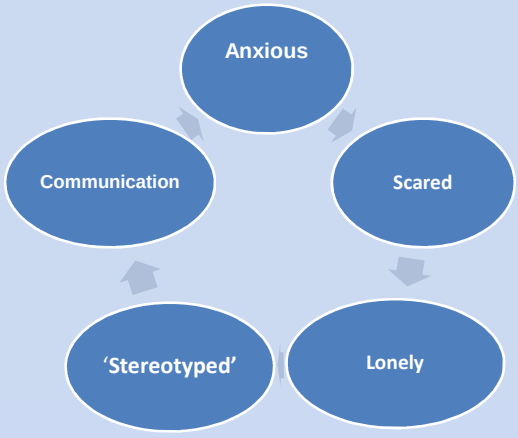

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Families with genetic conditions/Depends where you live?

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EGSP Team

If we go back to June the team was formed of:

- **Geneticist Lead Consultant - Dr Denise Williams**
- **Project Manager-Heena Jabbar**
- **Administrator***
- **1 full time GC post (2 job share)***
- **1 Community Educator-Maarya Moden**
- **1 Clinical Educator –Dr Amal Muflahi**

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Role of the Genetic Counsellor

- Help patients to understand genetic disease, recurrence risks.
- Disease management options.
- Risks and benefits if testing.
- Non directive assistance in the decision making process.
- Unbiased information.
- Taking a family history
- Making a family pedigree.
- Obtaining information and notes from other hospitals that may benefit in diagnosis.

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Type of patients seen in the project

- Family history of an autosomal recessive condition
- Minority ethnic groups
- Consanguinity
- How to make a referral:
- Enhanced Genetics Services Project referral form (Yellow sheet)
- Letter to Clinical Genetics/EGSP
- Fax if urgent
- Cancer family history form: www.bwhct.nhs.uk/west-midlands-family-cancer-service

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How to refer to EGSP

- **HC professionals**
- **Genetic Consultants and Genetic Counsellors**
- **Self referrals**
- **Family referrals**
- **Reviewing notes of Patients previously seen in genetics.**

- **Genetics**
- **Consultants**
- **GP**
- **Midwives/ Antenatal screening**
- **Neonatal screening (for Haemoglobinopathies)**
- **Self referrals**

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Referral guide to genetics

Family history

- Individuals with a known or suspected genetic condition
- Family history of a known or suspected genetic condition
- Cancer (Referral guidelines: http://www.bwhct.nhs.uk/west-midlands-family-cancer-service)
- Dementia & cardiovascular disease(particularly if early onset) or unexplained sudden death

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Referral guide to genetics

- **Reproductive issues**
- **Consanguineous couples (couples who are blood relatives).**
- **Couples with a history of recurrent miscarriage/stillbirth/infant death.**
- **Fetal abnormality detected by scan/blood test in pregnancy.**
- **Diagnosis/death of a child with a known or suspected genetic condition.**
- **Ethnic groups with increased risk of certain genetic conditions (e.g. Tay Sachs).**

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Referral guide to genetics

Development issues:

- Genetic condition diagnosed by routine new born screening
- Developmental delay/learning difficulty (if not under specialist care)
- Children with dysmorphic features/congenital anomalies
- Deafness/ hearing impairment (early onset/unknown cause)
- Blindness/ vision impairment(early onset/unknown cause)

Note: <u>This is not an exhaustive list.</u>

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In conclusion

- The Enhanced Genetic Services Project (EGSP) has aimed to raise awareness of and access to genetic services by black and minority ethnic minority groups in Birmingham, particularly the Pakistani community.
- EGSP focused on identifying families with a history of Recessive conditions by providing them with information to enable couples and individuals to make informed reproductive choices.
- Birmingham has a significantly increased infant and perinatal mortality rate in comparison to England and Wales.
- The project will end in November 2013 however, the education strand will continue and the overall evaluation report will be available form March 2014.

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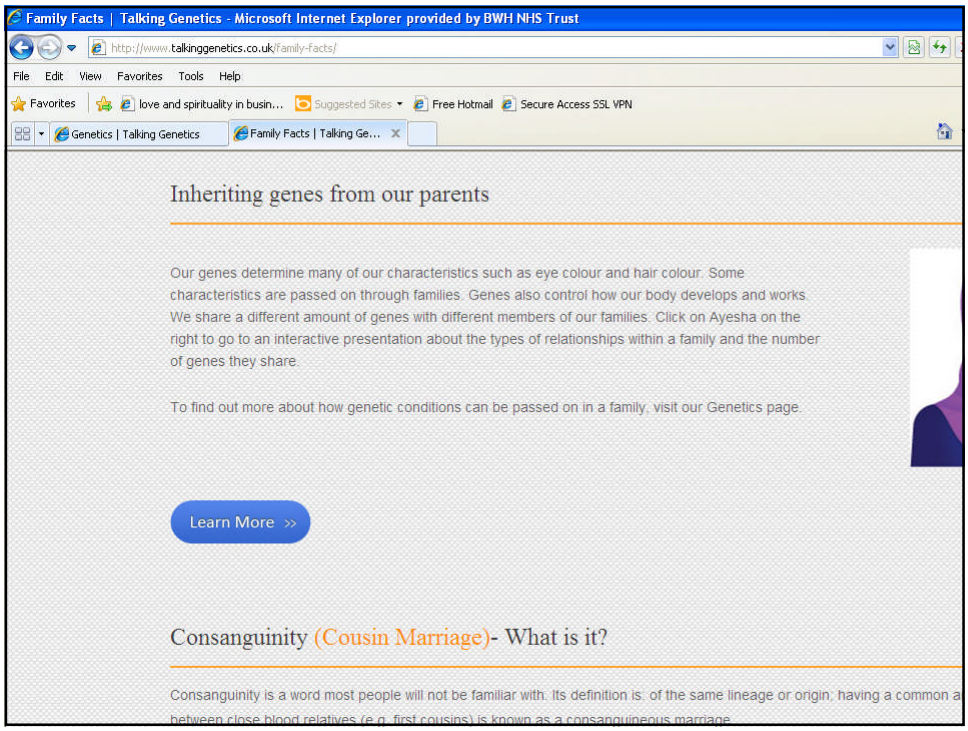

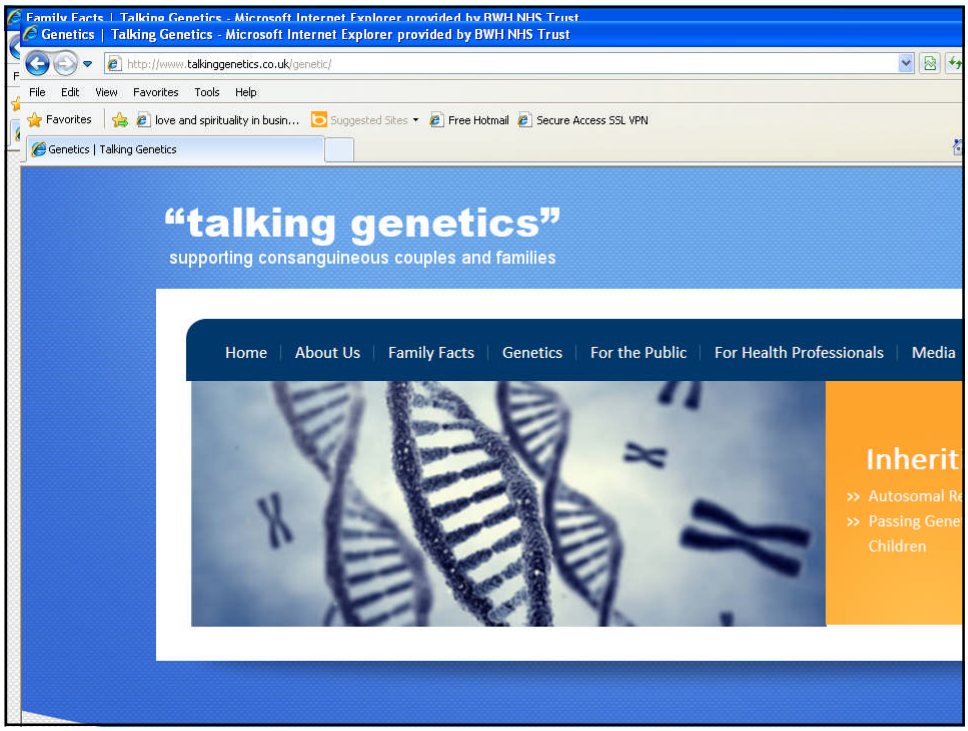
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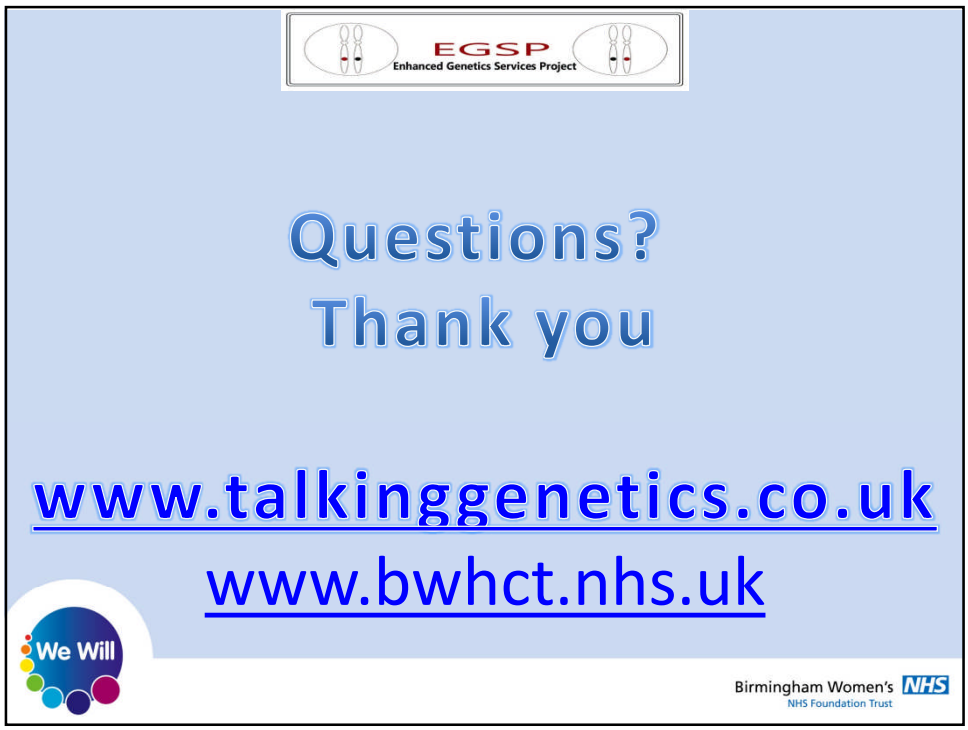
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Questions?
Thank you
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