

Consanguinity and Child Health: *Factsheet*

Tower Hamlets Joint Strategic Needs Assessment 2014-2015

UPDATED August 2014

Executive Summary

Consanguineous marriage is a union between couples related as second cousins or closer. Globally, 10.4% of the population are married to a biological relative or a progeny of such a relationship. While there are potential social, economic, and genetic advantages to consanguineous marriages, there is also a significant association between consanguinity and increased risk of child mortality, disability and other conditions linked to autosomal recessive inheritance. An analysis of the impact of consanguinity locally and of current service provision in terms of genetic testing and counselling was undertaken in response to reports that consanguinity may be a contributory factor to some cases of child disability and death.

The findings of the review are that there appears to be a substantially higher prevalence of consanguinity in the population of children with developmental delay in Tower Hamlets known to the Child Development Clinic (18-25% of cases) than in the general population (8% of births). This could represent an over-representation of consanguinity in the population of children with developmental delay but it could also reflect confounding factors such as ethnicity or socioeconomic status and further investigation of this is required. In the children with related parents known to the Child Development Clinic, 80% speak Bengali or Sylheti at home which has potential implications for service planning.

A number of areas in the UK where there is a high prevalence of consanguinity have implemented interventions to respond to the increased genetic risk associated with consanguineous marriage and further work needs to be undertaken to consider if such an approach should be undertaken in Tower Hamlets. However, it is important to acknowledge that the challenge in tackling this potential risk can be complicated by cultural sensitivities, preconceptions and misconceptions around consanguinity.

Recommendations

1. Further data analysis is recommended as follows:
 - Monitor antenatal data to see if there is a trend over time in the proportion of consanguineous relationships leading to conception
 - Explore other variables and birth outcomes in the antenatal dataset e.g. ethnicity, to allow consideration of confounding variables
 - Complete the data analysis of the Child Development Team Clinic data to compare the relative proportions of children known to the service with autosomal recessive conditions in those with non-related parents and those with related parents and to identify the level of disability experienced by these children
2. Engage with the clinical genetics service at Great Ormond Street Hospital to discuss current service provision in terms of genetic testing and counseling for families
3. Liaise with other regions in England that have implemented an approach to responding to the increased genetic risk associated with consanguineous marriage
4. Work with community leaders to agree how to explore the public perspective of consanguinity and service needs in a culturally sensitive manner in Tower Hamlets
5. Consider holding a workshop with community / genetics / antenatal services to discuss and agree approach to discussing consanguinity and related recessive gene disorders
6. Work with the local community to develop a culturally sensitive strategy that addresses the impact of consanguinity and empowers families to make decisions about managing their own genetic risk

1. What is Consanguinity?

Consanguineous marriage is commonly defined as a union between couples related as second cousins or closer. Most forms of consanguineous union are legal in the UK (exceptions include relationships between certain second-degree relatives (e.g. uncle–niece, half-siblings) and incestuous relationships (parent–child, brother–sister). Globally, 10.4% of the population are married to a biological relative or a progeny of such a relationship.¹

A number of social and economic advantages of consanguineous marriage are have been identified including:²

- Assurance of marrying within the family and the strengthening of family and societal ties
- Assurance of knowing one’s spouse before marriage
- Reduced chances of maltreatment or desertion
- Simplified premarital negotiations, with conditions and arrangement agreed in late childhood or early teens
- Greater social compatibility of the bride with her husband’s family, in particular her mother-in-law who also is a relative
- Reduced dowry
- Maintenance of land holdings

However, a significant association has been consistently demonstrated between consanguinity and the risk of mortality and morbidity resulting from congenital defects arising from autosomal recessive inheritance. ³ The decline in overall infant mortality to very low rates in England has revealed the contribution of severe recessive disorders to childhood mortality and morbidity.⁴

Child Health Burden of Consanguinity⁵

Cousin marriage impacts almost exclusively on recessively inherited conditions and does not influence chromosomal abnormalities, sex-linked or dominantly inherited conditions. However, the range of autosomal recessive conditions is extremely wide and not clearly defined. A preliminary survey in Blackburn found that the incidence of autosomal recessive disorders in childhood was 12 times greater in the Asian population than in Caucasians with 83 different recessive disorders being identified.⁶ However, accurate estimates of the increased genetic risk associated with consanguinity are hampered by poor data availability.⁷ A prospective study in Birmingham reported that among a sample of over 2,000 North European babies the birth prevalence of all congenital disorders was 4.3% (with 0.28% being identified as possible recessive disorders), compared to 7.9% (with around 3% being recessives) among the 956 British Pakistani babies in the study. It is estimated that of around 2,300 children born annually in the UK with a severe recessive disorder at least 630 (30%) are from parents of Pakistani origin (who contribute just 3.4% of all births).⁸

Confounding factors

Non-genetic variables are known to influence childhood health including social conditions, maternal age and education, birth order, and birth intervals. However, mean maternal age at marriage and at first birth is generally lower in consanguineous unions and there also is evidence that women in consanguineous unions continue to bear

¹ Bittles and Black, 2010. The impact of consanguinity on neonatal and infant health. *Early Human Development* 86 (2010) 737–741

² Saggar, A.K. and Bittles, A.H. (2008) *Consanguinity and child health*. *Paediatrics and Child Health*, 18 (5) pp. 244-249

³ Bittles and Black (2010)

⁴ Aamra Darr (2010) HGSG Briefing Paper Consanguineous Marriage and Inherited Disorders

⁵ Bittles and Black (2010)

⁶ Khan et al 2010 *J Community Genet* June; 1(2): 73–81. Developing and evaluating a culturally appropriate genetic service for consanguineous South Asian families

⁷ Salway et al (2012). Responding to increased genetic risk associated with consanguineous marriage: A formative review of current service approaches in England. Available at <http://clahrc-sy.nihr.ac.uk/images/health%20inequalities/resources/Responding%20to%20increased%20genetic%20risk.pdf>

⁸ Darr et al, 2010

children at later ages. It is therefore extremely important to consider the following confounding variables when considering the impact of consanguinity on child health:

- Gender (disability prevalence higher in boys)
- Age (cohort effects)
- Socioeconomic status
- Maternal age and education
- Birth order and birth intervals
- Reproductive behavioural factors including longer reproductive span (younger maternal age and bear children up to later ages)

Rates of Foetal Loss

- The evidence is not clear to date of the relationship between consanguinity and foetal loss. The majority of studies do not indicate a higher rate of foetal loss for consanguineous couples but these studies tend to focus on losses later in pregnancy and losses due to genetic disorders / other causes may occur earlier in pregnancy.

Birth outcomes

- Stillbirths are in excess of 1.5% deaths at first cousin level
- Evidence is not clear on the relationship between consanguinity and birth weight

Deaths in neonatal period and infancy

- Excess of 1.1% deaths in first cousin progeny both in the neonatal period and in infancy but confounders are often not accounted for in studies

Birth defects

Congenital anomaly

- A recent study in Bradford found that consanguinity was associated with a doubling of risk for congenital anomaly and that 31% of all anomalies in children of Pakistani origin could be attributed to consanguinity. The authors conclude that consanguinity is a major risk factor for congenital anomaly.⁹

Deafness

- Increased incidence of both syndromic and non-syndromic deafness with consanguinity

Visual impairment

- Consanguinity-associated blindness is less frequent than deafness but retinitis pigmentosa and congenital cataracts have been associated with consanguinity

Congenital heart disease

- Elevated rates of consanguinity have been consistently reported for congenital heart defects such as atrial septal defects and ventricular septal defects.
- There are variable reports of other congenital cardiac abnormalities including transposition of the great arteries, coarctation, pulmonary atresia and Tetralogy of Fallot.

Other defects

- Neural tube defects are suggested to be more common but this could be related to confounding variables
- There may be a genetic explanation for an association between Down syndrome and consanguinity but there is insufficient evidence of this to date

Single gene autosomal recessive diseases

- Genetics suggest that consanguinity is much higher in rare metabolic conditions e.g. lysosomal storage disorders and cerebral lipidoses.

⁹ Sheridan et al, 2013 (in press) Lancet. Risk factors for congenital anomaly in a multiethnic birth cohort: an analysis of the Born in Bradford study

Blood disorders

- Increased prevalences of α - and β - thalassaemias, rare complex haemoglobinopathies and other haematological disorders are seen with consanguinity

Developmental delay

- Mild and severe intellectual and developmental disability present in higher prevalence in consanguineous unions

An over-emphasis on the contributory role of consanguinity alone to ill-health has led to misconceptions and caused unease and upset in communities which traditionally have favoured consanguineous marriage. It should be noted that consanguineous marriage is not restricted to specific religions or population groups¹⁰ and also that:

- Consanguinity facilitates expression of rare recessive disease genes but does not cause genetic disease.
- Consanguinity can have favourable as well as unfavourable biological effects.
- In populations which favour consanguineous marriage, the circle of family members who can act as successful tissue donors also is significantly extended.
- Many rare recessive disorders are transmitted by healthy parents who carry one gene variant for the disorder. The harmful recessive gene mutations tend to cluster within extended family groups, but even when both parents carry the same abnormal recessive gene, the chance of each pregnancy being affected by that condition is 1 in 4. Therefore, most babies born to cousin couples are healthy.

2. What is the Policy Context?

Legal Considerations

First cousin and other more remote categories of consanguineous marriage are permissible under civil legislation virtually throughout the world, with the notable exception of the USA (restrictive laws in 31 states).

National Policy

The Department of Health does not provide general guidance to the public on cousin marriage, but it supports NHS initiatives among communities with a higher prevalence of cousin marriage on a number of issues. In particular, DH supports the need to work with communities to increase the understanding of genetic risk and raise awareness of the availability of genetics services that can provide advice and support for at risk families.¹¹

The Department of Health published the *Implementation Plan for Reducing Health Inequalities in Infant Mortality: A Good Practice Guide* in 2007.¹² This provides details on how genetic screening and counselling services play an important part in this work. Specifically, it highlights the work of two DH-funded projects on how to provide appropriate genetic services and support to communities that practise cousin marriage.¹³

Other national policy documents include guidance that is relevant to this issue without specific mention of consanguinity. These include:

1) *The National Service Framework (NSF) for Children, Young People and Maternity Services Standard 8*¹⁴: Marker of Good Practice 3: Early identification and intervention are provided through clinical diagnosis and the *Framework for the Assessment of Children in Need and their Families*. Interventions support optimal physical, cognitive and social development, and are provided as early as possible with minimum waiting times.

¹⁰ Bittles and Black (2010)

¹¹ Darr (2010) HGSG Briefing Paper Consanguineous Marriage and Inherited Disorders

¹² <http://www.chimat.org.uk/resource/item.aspx?RID=58176>

¹³ Darr (2010) HGSG Briefing Paper Consanguineous Marriage and Inherited Disorders

¹⁴ The National Service Framework (NSF) for Children, Young People and Maternity Services Standard 8 (2004)

http://www.dh.gov.uk/prod_consum_dh/groups/dh_digitalassets/@dh/@en/documents/digitalasset/dh_4090556.pdf

This is of particular relevance as there is the potential that many children are known to have developmental delay, possibly caused by an autosomal recessive condition, but without a definitive clinical diagnosis. Genetic testing, particularly in the context of parental consanguinity, could enable earlier diagnosis and intervention and informed future reproductive behaviour decision-making.

2) *Maternity Matters* (2007):

Policy commitment to maternity services:

“1.2 The aim of health reform in England is “to develop a patient-led NHS that uses available resources as effectively and fairly as possible to promote health, reduce health inequalities and deliver the best and safest healthcare”. For maternity services this means providing high quality, safe and accessible services that are both women-focused and family-centred. Services should be accessible to all women and be designed to take full account of their individual needs, including different language, cultural, religious and social needs or particular needs related to disability, including learning disability.”

NICE Clinical Guideline 62 covers Antenatal care states that areas outside the remit of the guideline include when there is a family history of genetic disorder, or women who have had recurrent miscarriage, a stillbirth or neonatal death, a small for gestational age baby, or a baby with a congenital anomaly.¹⁵ There is no explicit mention of consanguinity and the potential impact of consanguinity is largely outside the remit of the document.

Community genetics services in low- and middle-income countries: Report of a WHO Consultation (WHO 2011)¹⁶

This WHO consultation considered genetics services in a different setting to the UK, however some of the issues and recommendations are relevant including:

5.1.1.3 Pre-conception care and family planning

Pre-conception care is known to improve pregnancy outcome. Genetic components of preconception care include: detection of genetic risks through family history; addressing the issue of consanguinity if relevant; explaining programmes of prevention of congenital disorders and genetic diseases that exist in the community; and genetics counselling as appropriate.

5.3.3. Education in genetics

Education in genetics should be provided to all health professionals, policy-makers, and the public and needs to include topics such as: consanguinity; prenatal and newborn screening and detection of signs of possible genetic conditions common in the area; prevention and care programmes that take place in the community; and ethical, legal and social issues. Education in genetics is required to implement efficiently community genetics services in primary health care.

7.3. Ethical standards for community genetics services

The principal ethical standards for community genetics services are based on maximizing benefit, minimizing harm, respecting privacy and autonomy and ensuring equity and can be summarized as follows:

- fair allocation of public resources so that genetics services reach all those in need
- freedom of choice in all matters relevant to community genetics services; the necessity of voluntary approaches in services, including approaches to testing and treatment; the avoidance of coercion by government, society, or health professionals
- respect for human diversity, religious, cultural and social beliefs and psychosocial wellbeing
- education in genetics for the public, medical and other health professionals, teachers, clergy and others
- provision of ongoing quality control of services, including laboratory procedures

¹⁵ NICE CG62: Antenatal care (2008) <http://www.nice.org.uk/nicemedia/live/11947/40145/40145.pdf>

¹⁶ http://whqlibdoc.who.int/publications/2011/9789241501149_eng.pdf

Local policies for Tower Hamlets

The Tower Hamlets Children and Families Plan 2012-2015 includes that work needs to be undertaken to investigate the local prevalence of consanguinity and its impact on child health to inform an assessment of need for genetic counselling and wider awareness raising in affected communities.

3. What is the local picture?

Data on consanguinity are collected by a number of services in Tower Hamlets but these data have not routinely been collated or analysed at the population level. The Tower Hamlets Infant Mortality JSNA factsheet and Children and Families Plan 2012-2015 have both highlighted the need for more local information on consanguinity and an analysis of the impact of consanguinity on infant mortality and morbidity.

Population and births in Tower Hamlets

The borough has seen a significant increase in the number of young children since 2001, with a 19.8% increase in the number of residents aged under 10 from 28,542 in 2001 to 34,200 in 2011 (an increase of 5,658 children)¹⁷. 4,565 children were born to Tower Hamlets residents in 2010. The birth rate in Tower Hamlets is 66.2 live births per 1,000 females aged 15 to 44. The birth rate in Tower Hamlets is lower than the average in London (72.1), but about the same as England (65.5). In the 5 years between 2009/10 and 2014/15 the rate is projected to increase by 5.3% (235 additional births); in the subsequent 5 years between 2014/15 to 2019/20 the rate is projected to increase by 2.2% (100 additional births).

Ethnicity

There are a number of reasons why the ethnicity of mothers in a local area may have an influence on the needs which the services provided must meet. These include that certain conditions are known to be more common in particular ethnic groups; mothers or their families who have recently moved to the UK may have difficulties reading or speaking English; and different cultural norms may exist.¹⁸ In 2009, 41.1% of all women aged 16 to 59 years in Tower Hamlets PCT were from black and minority ethnic (BME) groups.

The table below shows the distribution of deliveries by mothers' ethnicity in 2010-11¹⁹:

	% of deliveries by ethnicity of mother						
	White	Asian / Asian British	Black / Black British	Chinese / Other	Mixed	Not known	Not stated
Tower Hamlets	23.0	49.9	7.3	6.5	1.3	1.6	10.4

The Asian and Asian British mothers in Tower Hamlets are mostly of Bangladeshi origin.. The average birth rate per 1,000 for Bangladeshi women aged 15 to 44 is 1.52 times the average for all women and Bangladeshi mothers also tend to be younger than non-Bangladeshi mothers.

Consanguinity in Tower Hamlets

Data on consanguinity is limited both nationally and locally and there is only fragmentary information on the prevalence of consanguineous marriage in Bangladesh. However, consanguinity does appear to be quite common in the UK Bangladeshi community but specific data for this is not available.²⁰

¹⁷ Census data, 2011

¹⁸ Child and Maternal Health Observatory

¹⁹ Hospital Episode Statistics (HES), The NHS Information Centre for health and social care

²⁰ Bittles (2010)

1) Antenatal data

Antenatal data from women booked in the Barts Health London site (of whom 91% are Tower Hamlets residents) indicates that there were 4641 live births in 2011/12 and 385 women had documented consanguinity. Therefore an estimated **8.2%** births resulted from a consanguineous union in 2011/12.

However, limitations of this data include that some women may not have been asked for this information which would lead to a potential underestimation of consanguinity, a union with any blood relation is included (not limited to e.g. first cousin), there were no further details collected on the nature of the relationship, and the level of data accuracy is not known.

2) Child Development Team (CDT) clinic data

The CDT clinic provides an assessment and co-ordination service for children with multiple disabilities and their families when 2 or more areas of developmental delay are identified in the child. All children resident in Tower Hamlets who meet the referral criteria should be referred to this single service. Local data from the CDT clinic were collated and described below including data on consanguinity which is recorded during the initial assessment of children.

514 children were identified as seen for the first time in the CDT clinic between 1/1/08 and 14/3/13.

	All children seen in CDT clinic 08–1/3/13	Related parents	Non-related parents	Unknown consanguinity status
Total number (%)	514	91/355 (25.6% of children whom status was known)	264/355 (74.4% of children whom status was known)	159/514 (30.9%)
% male	68.3% (347/508)	68.1% (62/91)	67.3% (175/260)	110/157 (70.1%)
% Bangladeshi ethnicity	47.7% (235/493)	76.9% (70/91)	40.2% (103/256)	43.2% (63/146)
% speak Bengali or Sylheti at home (+/- English)	53.2% (209/393)	80.5% (66/82)	41.8% (95/229)	57.1% (48/84)

There are a number of limitations with this data. Relating to ethnicity coding, 44/493 patients with a recorded ethnicity were identified as Asian – any other Asian background. It is estimated that a large proportion of these will also be Bangladeshi and that this is a coding issue.

A second and greater issue is that consanguinity status could not be ascertained in 30.9% of cases. There may be a bias introduced by this as parents from certain ethnic backgrounds may be more likely to be asked if they are related. However, if we assumed the extreme case of none of the unknown group being from a consanguineous union then the proportion of children seen in the CDT clinic with related parents would still be **17.7%** (91/514) which is still more than double the rate identified by the antenatal data. However, if we only include those for whom consanguinity status is known, **25.6%** of children seen in the CDT clinic between 2008 and 2013 have parents who are related.

Relationship of parents of children known to be from a consanguineous union

61 = first cousins (67.0%)

5 = second cousins (5.5%)

10 = distant cousins (includes third cousins) (11.0%)

15 = unknown relationship (16.5%)

Diagnoses of children known to be from a consanguineous union

We could only ascertain a definitive diagnosis in 53.8% (n=49) of the children (excluding non-specific diagnoses of learning disability and developmental delay). A wide spectrum of conditions were diagnosed including cerebral palsy, autism, seizure disorders, Down's syndrome, and problems associated with ex-prematurity. A number of potentially autosomal recessive conditions were diagnosed including: Arnold- chiari malformation, Pompe disease, Poland-möebius syndrome, Pierre Robin sequence, Aicardi syndrome, Alagille syndrome, Bardet-biedl syndrome and other identified chromosomal abnormalities.

The 4 most common diagnoses in all 3 groups (related parents, non-related parents and unknown if related) were autism spectrum disorder, Down's syndrome, problems associated with ex-prematurity and cerebral palsy.

3) Birth weight

There is a high prevalence of gestational diabetes (9%) in the borough and a 2005/06 audit found that 81.7% of women with gestational diabetes were Bangladeshi. Diabetes in pregnancy is associated with a number of poor foetal and maternal health outcomes and early detection and management, together with on-going lifestyle modification will offer benefits to both mother and baby before, during and after pregnancy. A high proportion of the babies born in the borough have a low birth weight which also increases the risk of type 2 diabetes, as well as cardiovascular disease in later life. However, despite a low birth rate levels of infant mortality in the borough are not significantly different to the rest of London and England. [see JSNA factsheet on Infant Health]

4) Deaths in neonatal period and infancy

During the period 2004-10, the largest cause of death for infants over 28 days was congenital malformations, deformations and chromosomal conditions.²¹ There are approximately 25 child deaths per year in Tower Hamlets. A recent audit of the Child Death Overview Panel (CDOP) data found that consanguinity was identified as a possible contributory factor to the death in roughly **25%** of cases.

5) Sensory impairment

Previous research in Tower Hamlets children could not attribute high rates of deafness in Bangladeshi children to consanguinity alone [reference check Bajaj et al]

6) Disability

Measuring disability in childhood is difficult, because the notion of disability is multi-dimensional, dynamic and contested. Definitions vary across different settings. Most robust estimates and local data suggest that there are approximately 2,000 children and young people aged 0-19 with a disability in Tower Hamlets.

4. What are the effective interventions?

In 2012, a review of approaches to responding to increased genetic risk associated with consanguineous marriage was undertaken.²² Four areas were identified in England where significant initiatives were underway - Birmingham, Bradford, Walsall and Blackburn with Darwen. The key findings of this review were:

Structure of interventions

- Interventions across the 4 sites varied but included:
 - (1) family-centred genetics services for at risk individuals and families
 - (2) enhancement of competence of health professionals
 - (3) community level genetic literacy

²¹ Infant Health JSNA

²² Salway et al (2012) Responding to increased genetic risk associated with consanguineous marriage: A formative review of current service approaches in England. Available at <http://clahrc-sy.nihr.ac.uk/images/health%20inequalities/resources/Responding%20to%20increased%20genetic%20risk.pdf>

- 3 sites successfully developed a family-centred genetic service intervention focused on 'at risk' consanguineous families using a model of retrospective case review to identify individuals who might benefit from an enhanced offer, supplemented by establishment of prospective referral mechanisms
- Interventions at community level to enhance genetic literacy took a variety of forms.

Challenges

- Recruiting suitable staff with both genetics counselling and cultural competence skills
- Achieving cascade counselling and testing of extended family members
- Issues of persistent mistrust among community members and uncertainty about whether and how to engage with religious leaders were prominent
- Engagement of GPs was felt to be important but had not been successful indicating the need for new engagement strategies
- Efforts to engage and equip the wider healthcare workforce primarily consisted of voluntary face-to-face training events. Though largely well-received, these were not always linked to other aspects of service provision and lacked clear messages regarding whether and how practitioners' behaviours should change.
- Potential synergies between the three strands of recommended intervention were recognised, but there are conflicting views regarding the appropriateness of some elements of intervention.
- Cross-cutting challenges included: how to identify appropriate outcome indicators and measure success; how to ensure interventions are sustainable when the numbers of staff involved are very small and resources are vulnerable; how to develop a Community of Practice and avoid duplication of effort in different places; how to embed formal evaluation into future initiatives; and how to gain community involvement and trust.

Evaluation

- Limited formal evaluation
- Service users largely had high levels of satisfaction

Overall

- A consensus is emerging around the key elements of an appropriate service response to the issue of customary consanguineous marriage and genetic risk.
- However, important questions remain regarding how such a response can be operationalised effectively and cost effectively in practice

Blackburn and Darwen as an example:

Programme

- Provided a genetic service accessible to consanguineous families from the South Asian community with a child affected by an autosomal recessive disorder
- Provided information on genetic risk along with the offer of genetic testing for members of the extended family to identify gene carriers and facilitate informed reproductive choices.
- Employed an Urdu-speaking health visitor to establish a community-based, hospital-linked genetic service
- Offered local paediatric and regional genetic services to parents who had an affected child and their relatives

Evaluation

- High uptake of service (95% of index parents and 92% of relatives to whom it was offered)
- High uptake of carrier testing (94% of relatives to whom it was offered)
- Eight requests for prenatal diagnosis were made during the course of the service development.
- Many individuals stated they would consider genetic risk when making future marriage and reproductive plans. Input from a health care worker from the same ethnic background who provided information in their own language was highly valued.

Conclusion

Family orientated genetic services for ethnic groups practicing consanguinity can be acceptable and effective when provided in a culturally appropriate manner.

5. What is being done locally to address this issue?

The Child Development Clinic provides an assessment and co-ordination service for children with multiple disabilities and their families where there are 2 or more areas of developmental delay identified in the child. All children who meet the referral criteria should be referred to this single service. A clinical decision is made on an individual basis of when to refer to the Clinical Genetics Service at Great Ormond Street Hospital and genetic tests and genetic counseling are carried out as deemed appropriate.

There is not a programme of pro-active awareness-raising about consanguinity in the Bengali community in Tower Hamlets.

6. What evidence is there that we are making a difference?

We do not have information on this currently.

Impact on Public Health Outcomes Framework indicators

2.5 Child development at 2-2.5 years

Earlier identification and diagnosis of autosomal recessive conditions may enable earlier intervention and support to improve development.

4.1 Infant mortality

Genetic counselling for families in the case of a severe autosomal recessive condition being diagnosed in one child may influence subsequent reproductive behaviour. However, the aim of community awareness-raising is not to necessarily influence decisions but to give families appropriate information to enable them to make an informed decision.

7. What is the perspective of the public on support available to them?

The public perspective of consanguineous relationships and genetic sequelae needs to be explored in a culturally sensitive manner in Tower Hamlets, particularly in the Bangladeshi population. Future work should include liaising with the Bangladeshi advisors and Sylheti / Bengali speaking staff to decide how best to approach this.

8. What more do we need to know?

- Existing data sources need to be explored fully to ascertain the burden of consanguinity on child health in Tower Hamlets including antenatal data, and thalassaemia clinic data
- The work with the CDT clinic data need to be analysed further in relation to diagnosis, genetics referrals and the level of developmental delay experienced
- The perspectives of the community and service users need to be sensitively explored

9. What are the priorities for improvement over the next 5 years?

1. Further data analysis to be undertaken, including:
 - Monitor antenatal data over time to see if there is a trend over time in the proportion of consanguineous relationships leading to conception
 - Explore other variables and birth outcomes in the antenatal dataset e.g. ethnicity, and to allow consideration of confounding variables
 - Complete the data analysis of the Child Development Team Clinic data to compare the relative proportions of children known to the service with autosomal recessive conditions in those with non-related parents and those with related parents and to identify the level of disability experienced by these children
2. Engage with the clinical genetics service at Great Ormond Street Hospital to discuss current service provision in

terms of genetic testing and counseling for families

3. Liaise with other regions in England that have implemented an approach to responding to the increased genetic risk associated with consanguineous marriage
4. Work with community leaders to agree how to explore the public perspective of consanguinity and service needs in a culturally sensitive manner in Tower Hamlets
5. Consider holding a workshop with community / genetics / antenatal services to discuss and agree approach to discussing consanguinity and related recessive gene disorders
6. Work with the local community to develop a culturally sensitive strategy that addresses the impact of consanguinity and empowers families to make decisions about managing their own genetic risk

10. Contacts / Stakeholder Involvement

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