OLDHAM HEALTH AND WELLBEING BOARD

Reducing the impact of consanguinity on child health in Oldham


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What is required from Health and Wellbeing Board?

A systematic response to the challenges arising from consanguinity in Oldham is necessary.

This includes the provision of adequate genetic counselling for at risk families in Oldham, training and workforce development for health and social care professionals to provide information and advice to clients, increasing the uptake of universal services, and action to raise knowledge in communities affected by consanguinity with regard to the associated risks, including the adoption of available communication tools for professionals working with affected families and communities.

The Health and Wellbeing Board is asked to delegate the implementation of this systematic response to the Integrated Commissioning Partnership in Oldham and to request regular updates on progress.

1.0 Background

1.1 Oldham’s Local Safeguarding Children’s Board, and the NE Sector Child Death Overview Panel identified concern regarding the apparently higher than national mortality and morbidity rates in relation to autosomal recessive disorders linked to consanguineous relationships in Oldham.

1.2 Partners therefore wished to consider the effects of consanguinity in Oldham more fully, and set out a planned response to reducing the impact of consanguinity related health issues in childhood that could be adopted as a shared approach within the borough.

1.3 This report describes current thinking from the literature in relation to consanguinity, outlines key risks and issues in relation to consanguinity in the borough, and proposes a number of actions to improve outcomes for families and communities.
2.0 What is consanguinity, and how common is it?

2.1 Consanguineous relationships are those between blood relatives, usually defined as those between first cousins or other close family members\(^1\). Marriage between biological relatives, in particular first cousins, is widely popular and with some exceptions (the USA in particular) is broadly permissible in law in most parts of the world. Over 1000 million people live in countries where between 20 and 50% of unions are between couples related as second cousins or closer\(^2\). The most recent consanguinity estimate indicates that around 10.4% of the world’s population are either married to a biological relative, or are the progeny of a consanguineous union\(^3\).

2.2 The choice of a marriage partner is strongly influenced by geography, ethnicity, religion, education, social status and political beliefs. Virtually all traditional societies are divided into long-established communities, with limited inter-community marriage, including industrialised western societies which show similar, if less pronounced divisions.

2.3 In societies with large immigrant communities, recent arrivals typically marry within their ethnic and or religious community during the first and second post-immigration generations. Although a consideration, choice of partner is not predicated by the likely health effects upon future offspring.

2.4 Consanguinity in England is linked to certain cultural norms, in particular to individuals from certain south-east Asian communities. Around 55% of known consanguineous marriages in the UK are found within those from a Pakistani background\(^1\) although it is known that consanguineous unions also exist to some extent throughout all populations. Of interest is that consanguineous unions were commonplace and valued in most western countries until the mid-nineteenth century. Notable examples are Albert Einstein and Charles Darwin both of whom married first cousins.

2.5 The decline in consanguineous marriage in Western Europe has been attributed to perceptions of risk relating to the likelihood of major physical and or intellectual disability in offspring, but is at least as likely to be related to wider social change, including maternal employment and education.

3.0 What is the health risks associated with Consanguinity?

3.1 Each parent passes on one copy of their genes to their children. Most people carry one or two gene variants for a recessive disorder but remain unaffected. If their partner also carries this recessive gene, their children may be affected. Closely related couples are more likely to share the same recessive genes\(^1\).

3.2 If both parents are healthy (perhaps unknown) carriers for a recessive disorder, there are three possible outcomes for each of their children:

- A 1 in 4 (25%) chance that the child could be **affected** by a recessive disorder.
- A 1 in 2 (50%) chance that the child could be a **healthy carrier**.
- A 1 in 4 (25%) chance that the child could have 2 normal gene copies and therefore would **not be a carrier or affected**.

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With each pregnancy the odds remain the same. Just because one child has already been affected, the odds do not reduce for any subsequent pregnancy.4

3.3 A recent and well received multi-ethnic birth cohort study5 undertaken in Bradford has summarised the risk relating to the likelihood of the children of consanguineous unions having increased risk of congenital anomaly. Congenital anomalies are a broad term, with not all cases being related to recessive disorders. There are three recognised causes of congenital anomalies:

3.3.1 *Genetic* including chromosomal aberrations (e.g. Down syndrome) and Mendelian single-gene defects (e.g. achondroplasia or Holt-Oram syndrome). The proportion of genetic origin is estimated about 25% of total congenital abnormalities. Two conditions are thought to contribute to higher total prevalence of congenital abnormalities with genetic origin: (i) women giving birth after 35 years of age and (ii) high rate of consanguineous marriages.

3.3.2 *Environmental* eg infectious diseases, maternal diseases, teratogenic drugs, alcohol, smoking and environmental pollutants. The proportion of environmental origin may be about 15% of total congenital abnormalities.

3.3.3 *Complex* (multifactorial) origin caused by gene-environmental interaction when there may be predisposition to an environmental ‘trigger’. Most common congenital abnormalities (such as isolated neural-tube defects, orofacial clefts, cardiovascular malformations, congenital pyloric stenosis, congenital dislocation of the hip, undescended testis, hypospadias, etc) belong to this etiological group. The proportion of complex origin is estimated about 60% of total congenital abnormalities, although it is less clear.

The first of these (genetic) is of particular interest in consanguinity as it relates most closely to recessive disorders, however the latter (complex) is also of interest as aetiology is unclear.

3.4 The Bradford study was not able to exclude all non-genetic related risk factors completely (due to complexities in establishing causal relationships for some disorders), however it offers a useful estimation of risk as follows:

3.4.1 Of the 11,396 cases where data was available 18% (n=2013) were off-spring of first cousins, the majority being to parents of Pakistani origin (n=1922).

3.4.2 The number of births where there was a congenital anomaly was 386 (19%), and of these, two thirds (n=233) were from mothers from a Pakistani background. It is important to note that risk is not uniform - some extended families are at greater risk than others depending on their history of recessive disorders.6

3.4.3 Births to Pakistani mothers made up 45% (5127 births) of the total cohort in Bradford. The data could not account for the effects of ‘birederi’, ie the effect of longstanding traditions of consanguinity within substructures of the Pakistani population particularly as information relating to lineage may not always be known within families.

3.4.4 Deprivation showed no increased association, although a similar increase in risk to that of consanguinity is noted for mothers of white British origin who were older than 34 years (multivariate RR 1.83, 95% CI 1.14—3.00). Maternal education to degree level was found to be protective (RR 0.53, 95% CI 0.38—0.75), irrespective of ethnic origin.

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3.4.5 The overall risk of having a child with a congenital anomaly was 3%, with the risk for a parent from a Pakistani origin in a consanguineous relationship (compared to those of white British origin) calculated as 1.96 (95% CI 1.56—2.46), i.e. nearly double the risk.

4 The situation in Oldham

4.1 In order to obtain accurate figures for Oldham, a similar analysis to that undertaken in Bradford would be required but has not been undertaken to date. However from data on the local population and national and research based estimates it is clear that for the Pakistani Community at least (10.1% of Oldham’s population) there is likely to be increased risk for children born to parents from this community who are in a consanguineous union.

4.2 Oldham has a significant proportion of people from populations that favour cousin marriages. 22.5% of the Oldham population is from an ethnic minority group, including 10.1% Pakistani and 7.3% Bangladeshi.

4.3 In 2010 a modelling exercise estimated consanguinity related issues for local areas, and Oldham was placed 8th out of the top 20 boroughs for the proportion of parents estimated to be related. It also suggested that:

4.3.1 Although ethnic minorities make up 22.5% of the current population in Oldham, 30% (n=909) of total births in any year (n= approx. 3,000) in Oldham were classed as being from a BME background, reflecting the relatively higher birth rate amongst this population group.

4.3.2 Of births to mothers from a BME background (n=approx. 900) 12% (n=108) of births to mothers from a BME background were estimated to be from parents who were related, however the majority of infants would not have been affected.

4.3.3 A broad estimate would suggest around 20 births annually in Oldham with consanguinity related disorder, of whom around eleven each year would live with a chronic disability and the other children may not live beyond the first year of life.

4.4 Estimates suggests that in Oldham BME communities appear to experience 35%-54.5% more infant deaths in comparison to the non BME population, with some of this being related to consanguinity, although a large proportion are also likely to be linked to other factors such as relatively poor maternal health (eg higher rates of gestational diabetes), and reduced access to pre-conceptual and antenatal care.

5 What Works to reduce consanguinity related health problems.

5.1 Expert groups agree that consanguineous marriage is an integral part of cultural and social life. In addition, attempts to discourage consanguineous marriage are considered inappropriate and undesirable, and unlikely to succeed.

5.2 Whilst consensus is emerging relating to the key elements of a response, there is an absence of rigorous evidence. A recent review presented lessons learned and insights gained, and suggested a three stranded multi-professional approach:

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i. **Family centred genetics services** for at risk families, including identification of affected child(ren), family tracing and proactive offer of counselling and testing. Affected families are known to be at risk, thus preventive interventions can be targeted, and affected families often act as community advocates.

ii. **Training and workforce development**, to enhance competence and confidence of health professionals in relation to how to support affected families and provide information and advice, including the development of effective communication tools which can be used by professionals to communicate risk.

iii. **Community activity** to raise genetic literacy and encourage the wider uptake of universal and targeted services, particularly prenatal and antenatal care.

NB The importance of *integrated coordinated action across all three recommended areas* is highlighted.

6 **Local issues**

6.1 At present the allocation of resources for genetic services across Greater Manchester is not adjusted for areas at high risk of inherited disorders. Oldham receives services from Manchester Regional Genetics Service, but this is limited, clinical in nature rather than delivered within communities, and as such additional support is likely to be effective.

6.2 Discussing genetic risk information across cultural and language barriers is complex. Poor understanding, fear and stigma can act as barriers to uptake of standard genetics services amongst Pakistani families even where serious conditions have been identified.

6.3 Consequently many families remain unaware of the existing genetic services (genetic counselling, carrier testing) or available options for reducing their genetic risk (choice of partner, restriction of family size, prenatal diagnosis).

7 **Achieving Outcomes from the approach**

7.1 Reductions in infant mortality and childhood disability at population level will not be easily evidenced in the short to medium term. The primary rationale must be one of tackling inequality and empowering individuals with information to make informed choices.

7.2 The assumption that investment in interventions will be more than compensated by the cost savings achieved via some affected births being averted is plausible, but may only be seen over the longer term. However, averting just one affected case is likely to offer significant financial, social and personal benefits in the long-term.

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8 Summary of Proposed Actions

A systematic response to the challenges arising from consanguinity in Oldham is necessary. This is the provision of adequate genetic counselling for at risk families in Oldham, training for health and social care professionals to provide information and advice to clients, increasing the uptake of universal services, and action to raise knowledge in communities affected by consanguinity with regard to the associated risks.

The health and Wellbeing Board is asked to delegate the implementation of this systematic response to the Integrated Commissioning Partnership in Oldham and to request regular updates on progress. The following are itemised actions:

8.1 **Review genetics service provision** for at risk individuals and families, including identification of affected child(ren), family tracing and proactive offer of counselling and testing. Consider additional capacity by way of a community practitioner with expert knowledge of genetics to provide dedicated support and advice to affected families and work within affected communities to encourage community advocacy relating to consanguinity.

8.2 **Make training available** for primary care and community practitioners to increase confidence in discussing consanguinity related issues. In addition, lobby for the incorporation of genetics in the context of consanguinity as part of training programmes for health professionals in Greater Manchester.

8.3 Commission community activities which will **raise genetic literacy**, and develop a supporting communications plan around consanguinity to disseminate best practice.

8.4 **Maximise the impact of universal prenatal, antenatal care and early years care** and services through reviewing specifications relating to these services, to encourage optimal uptake.