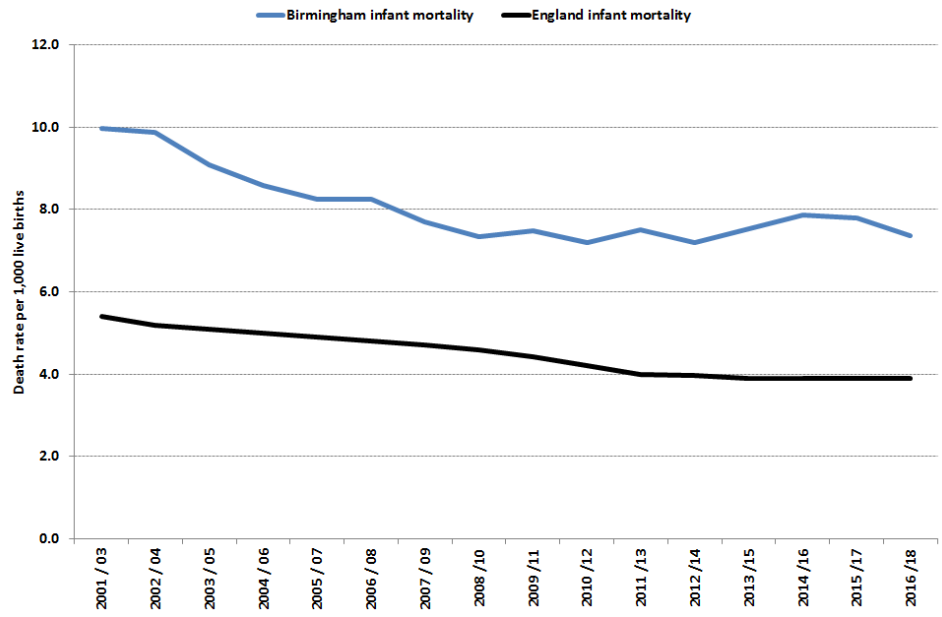
Briefing on Infant Mortality (deaths under the age of 1 year)

Introduction

Infant mortality is defined as death before the child reaches the age of one year. Infant mortality rate is the number of deaths under one year of age occurring among the live births in a given geographical area during a given year, per 1,000 live births. It represents a particularly distressing category of premature death and is an indicator of the general health of an entire population. The reason for the focus on infant mortality in Birmingham is due to Birmingham having consistently high rates that exceed that of England

Summary

In the three years between 2016 and 2018 there were 369 infant deaths in Birmingham (an average of 123 per year), representing an infant mortality rate of 7.4 per 1,000 live births. The infant mortality rate in Birmingham is significantly above the national average of 3.9 per 1,000 live births. Infant deaths account for around 88% of all deaths of children and young people in Birmingham (0-19).



Source: ONS Births and Deaths

Infant mortality rates have been declining steadily across the United Kingdon since the 2001/03 period. This has not been the trend in Birmingham where the infant mortality rate has not been consistently decreasing. In the period 2014-16 it rose to 7.9 per 1,000 and the period 2016 -18 it

decreased to 369 per 1,000. This is not a significant difference however; the rate is consistently above that of England.

The infant mortality rate consists of three components:

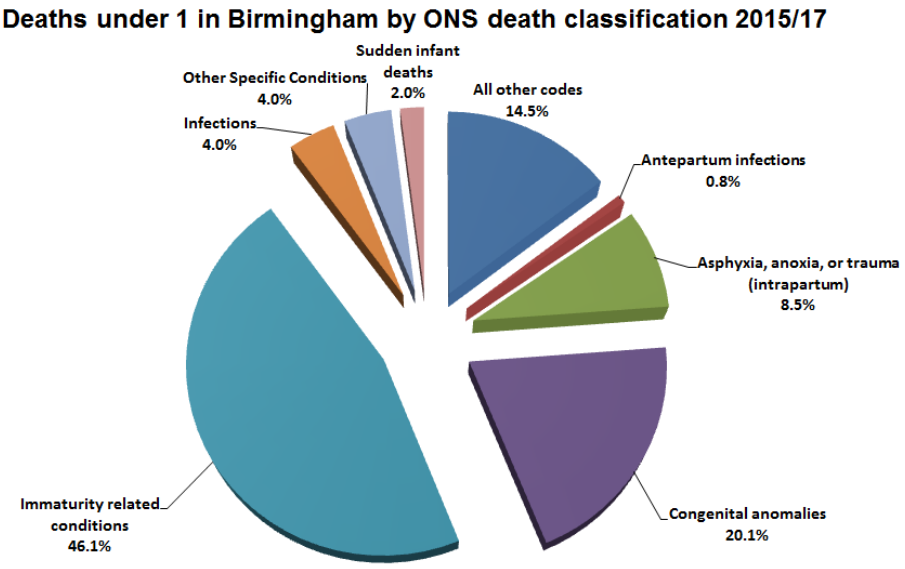
1. Early neonatal – the first 0 to 6 days after birth
2. Late neonatal – 7 to 28 days after birth
3. Post-neonatal morality rate: The number of infants who die between 28 days and less than one year

Influences on Health and Wellbeing

Infant mortality is an indicator of the general health of an entire population. The rate reflects the relationship between the wider determinants of population health such as economic, social and environmental conditions and the immediate causes of infant mortality. Deaths occurring during the first 28 days of life (neonatal period) are considered to reflect the health and care of both mother and newborn. There is a recognised correlation between higher infant mortality rates and deprivation. Reducing infant mortality overall and the gap between the richest and poorest groups are part of the Government's strategy for public health.

Consanguinity

Consanguineous marriage is a union between couples related as second cousins or closer. Globally, 10.4% of the population are married to a blood relative or a child of such a relationship.[i] 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.



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 Birmingham. 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.

A number of social and economic advantages of consanguineous marriage are have been identified including:[ii]

- 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. [iii]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.[iv]

Child Health Burden of Consanguinity[v]

Cousin marriage impacts almost exclusively on inherited conditions which are controlled by genes that are inherited from both parents i.e. recessive and does not influence chromosomal abnormalities that are sex-linked or dominantly inherited conditions. However, the range of autosomal (linked to a chromosome that is not a sex-linked) 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 North European with 83 different recessive disorders being identified.

[vi]However, accurate estimates of the increased genetic risk associated with consanguinity are hampered by poor data availability.[vii] 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).[viii]

Confounding factors that are 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 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.[ix]

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 lipoidoses.

Blood disorders

Increased prevalence's of α- and ß- thalassemias, 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.

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.

The Department of Health published the Implementation Plan for Reducing Health Inequalities in Infant Mortality: A Good Practice Guide in 2007.

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 814:

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.

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."

3) NICE Clinical Guideline 62

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. There is no explicit mention of consanguinity at its potential impact as it was largely outside the remit of this guideline.

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

This WHO consultation considered genetics in countries outside the UK, however, some of the issues and recommendations are relevant.

5) Pregnancy and early life: reducing stillbirth and infant death (2019):

A planning tool that examines factors that influence stillbirth and infant death at population level, rather than in individual clinical care.

6) NHS England Saving Babies' Lives: A care bundle for reducing stillbirth (2019):

Guidance that enables acute trusts to examine factors that influence stillbirth and develop actions to mitigate them.

Local policy

There is a systematic multiagency process for gathering data after every childhood death known as a Child Death Review. The Child Death Overview Panel gathers comprehensive information on the factors that contribute to a child death in order to make recommendations on appropriate changes in practice that are needed. In Birmingham, Child Death Reviews are carried out by the Birmingham Child Death Overview Board which reports to the Birmingham Safeguarding Children's Board.

The Birmingham and Solihull United Maternity and Newborn Partnership (BUMP) is a collection of local NHS Acute Trusts who provide maternity care, that have come together under one vision:

'To deliver a consistent world class holistic service that empowers women and families to make informed choices, enabling them to access high quality care from a range of providers that is most suited to their personal choice and clinical need.'

BUMP aims to introduce:

- A single point of access for all maternity referrals making sure you have access to the right care from day one, through your dedicated midwife
- Dedicated Community hubs – bringing midwifery and specialist care to convenient locations and

- A host of additional services, including online antenatal courses and much more.

The local situation

Local services that address this issue

The acute trusts provide an assessment service for parents at risk of genetic abnormalities due to recessive autosomal conditions. Once the results are known they are referred for appropriate genetic counselling if required.

There is not a programme of pro-active awareness raising about consanguinity across relevant communities in Birmingham.

What is the perspective of the public support available?

It is extremely important that cultural sensitivity is taken to ascertain what local communities feel about the support offered to them. Currently little is known and an evaluation is being planned to ascertain what communities feel about some of the themes covered within this report and what they consider can be implemented.

Recommendations

1) Undertake in-depth analysis to underpin the development of a Birmingham Infant Mortality Strategy

2) Review the provision of clinical genetics, genetic testing and counselling for families

3) Liaise with other regions in England such as Bradford, Sheffield and Tower Hamlets, that have implemented an approach to respond to the increased genetic risk associated with consanguineous marriage.

4) Work with community leaders to agree how to explore the public/ community perspective of consanguinity and service needs with a culturally sensitive approach

5) Hold a workshop with community/genetics/antenatal services to discuss and agree approach

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[i] Bittles and Black, 2010. The impact of consanguinity on neonatal and infant health. Early Human Development 86 (2010) 737–741
[ii] Saggar, A.K. and Bittles, A.H. (2008) Consanguinity and child health. Paediatrics and Child Health, 18 (5) pp. 244-249
[iii] Bittles and Black (2010)
[iv] Aamra Darr (2010) HGSG Briefing Paper Consanguineous Marriage and Inherited Disorders
[v] Bittles and Black (2010)
[vi] 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

[vii] 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://clahrcsy.nihr.ac.uk/images/health%20inequalities/resources/Responding%20to%20increased%20genetic%20risk.pdf

[viii] Darr et al, 2010

[ix] 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