NCMD
National Child Mortality Database

# NCMD Guidance for CDR Professionals and CDOPs on consanguinity

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## Acknowledgements

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## Introduction

The Children Act 2004, Working Together to Safeguard Children (2018) and the Child Death Review (CDR) Statutory and Operational Guidance together create a statutory requirement for Child Death Overview Panels (CDOPs) to review the deaths of all children in England who die before their 18th birthday. The purpose of this process is to understand how and why children die and to learn from these events to reduce the number of children who die in the future.

There are currently 58 CDOPs across England who provide multi-agency reviews of child deaths. Analysis of data provided by the National Child Mortality Database (NCMD) has shown that consanguinity is one of the most frequently recorded modifiable factors by CDOPs in their reviews. However, there is variability in how information is recorded and whether or not individual factors identified are considered to be contributory or modifiable.

CDOPs have the ability to influence commissioning of services, policy and decision-making in their areas and the data collected by the CDR process and analysed by NCMD is a vital tool to assist in improving services and access to them across the country.

Following the publication of the second NCMD annual report in June 2021, which highlighted that consanguinity was one of the most commonly recorded modifiable factors by CDOPs, the National Steering Group for Consanguinity and Genetic Risk challenged the robustness of the data due to the variability with which CDOPs considered consanguinity within their reviews and whether and how it was recorded. In addition, several conversations were already ongoing among CDOPs on how they could address this issue. This led to the setup of the task and finish group that compiled this document.

## Definitions

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| Consanguinity | Any couple related by blood to each other, also known as close relative marriage |
| Deleterious gene | A change in the DNA sequence of a gene that causes a person to have or be at risk of developing a certain genetic disorder or disease. |

## Purpose of the guidance

- To improve the consistency or recording of factors related to consanguinity
- To ensure that all CDOPs adopt the same approach when deciding whether issues related to consanguinity are recorded as modifiable or contributory
- To raise awareness and strengthen the response of CDR professionals in how they assess/consider deaths that may be related to consanguinity
- To improve the ability of CDR systems to engage with the agenda in a culturally competent and supportive manner to be better able to serve diverse communities who have experienced the death of a child that may be related to consanguinity
- To reduce unwarranted variation in practice and increase awareness among CDR professionals of how to discuss and record issues around consanguinity
- To ensure enhanced equity of access to appropriate, culturally competent, clinical genetic services for all families at increased genetic risk, to enable informed decision-making.

## Who is this guidance for?

Any professional working within the CDR system, particularly those chairing and attending Child Death Review Meetings (CDRMs) and members of Child Death Overview Panels in England.

## Close Relative Marriage

For consanguineous families with no family history of genetic conditions there is a slightly increased risk of having a child with a genetic disorder. However, once a family has a child with a genetic disorder that family is at increased risk as is their extended family.

In general, therefore, it is unacceptable to discourage close relative marriage in a blanket way. This is not appropriate given the level of risk since 90% of children born to consanguineous families will not be affected by a genetic condition[1].

In families where there is a known, autosomal recessive condition, genetic counsellors should give advice to families to consider arranging future marriages outside of the family to reduce the risk to the family. Action at community level may help people to understand and act on this advice; but this is only acceptable if information is balanced, non-stigmatising and non-directive.

## Information collection

CDRM and CDOP discussions must have all the information relating to each family's experience of care in order to review the child's death in a comprehensive and consistent way. In order to ensure accurate and complete information about the genetic condition of the child and the interaction between services and families the following actions should be taken:

- CDOP offices should set up arrangements with their local genetics service to perform a check on every child that dies of a likely genetic condition. For new-born babies, the check should be on the mother and baby.
- Where a child is known to genetic services, a reporting form, including the supplementary form relating to chromosomal, genetic and congenital anomalies, should be sent to the service to complete and/or ensure that clinical records are checked, and relevant genetic information is brought to the CDRM by the healthcare team for inclusion in the analysis form.

Stigma means families can be reluctant to disclose their relationship status and therefore when a death is discussed at a CDRM or CDOP professionals should ensure during their review that information is collected via the reporting form on the relationship status of every family not just those from certain ethnic groups. It is important to recognise that an anti-stigmatic process should be observed across the whole care pathway and discussions with families should take place in a culturally supportive manner.

## The Genetic Condition of the Child

Consanguineous families are at increased risk of being affected with autosomal recessive conditions. In autosomal recessive inheritance, a genetic condition occurs when the child inherits one mutated copy of a gene from each parent. If the child being reviewed is affected with a definite or likely autosomal recessive condition, it is likely to be related to consanguinity. It is important to know if autosomal dominant, mitochondrial, x-linked and chromosomal causes have been excluded for the affected child.

For consanguineous families with children who have no diagnosis when they die, if there is more than one affected child or adult in the family, professionals should consider that the condition might be related to consanguinity.

[1] Sheridan et al (2013), Teeuw et al (2010)

## Modifiable and Contributory Factors

Factors recorded on the analysis form in Domains A-D and graded as a 2 are considered to be contributory factors. Not all contributory factors will be modifiable. Modifiable factors are those which may have contributed to the death of the child, and which might, by means of a locally or nationally achievable intervention, be modified to reduce the risk of future deaths. The CDRM and CDOP meetings will record which are modifiable in the list of modifiable factors box on the analysis form.

- **Consanguinity itself should not be considered a modifiable factor**. Access to culturally competent genetic services is what is potentially modifiable.
- The focus should be on whether the *death* of the child with the genetic condition could have been avoided or future deaths from the same condition could be prevented in this family.
- For children with autosomal recessive or likely autosomal recessive conditions, that die of their condition, or from complications arising from their condition, consanguinity should be recorded on the analysis form as a contributory factor and graded as 2.
- For children with such conditions who die of something unrelated e.g. in a road traffic collision, consanguinity should be recorded on the analysis form and graded as 1.
- The flow chart included in Appendix A of this document should be followed to help decide on modifiability

It is important for the CDRM and CDOP to consider whether the family had all the information they needed to make an informed choice about their pregnancy.

## During the CDRM/CDOP review

Professionals should consider:

- Whether the family knew that they were at increased risk of having a child with a genetic condition. For example, was there information held within multi-agency systems to suggest that a child with a serious genetic condition might be born and if so, was this information shared with the family as part of their decision-making process? Were genetic services offered to the family in a culturally competent and accessible way as this may affect the uptake of services by families? For example: using an interpreting service, advocacy from people of the same culture / people with existing trusted relationships with the family.
- Was this the first affected pregnancy? If yes, then it is likely not modifiable.
- Has genetic testing been undertaken in the family to empower families with the option of informed reproductive decision making in future pregnancies for example the option of pre-natal diagnosis?
- Did the child receive appropriate investigations during life? A lack of investigation reduces the options for families in a future pregnancy.
- Is there evidence that the family has been referred to genetic services following the death of their child? If the child had a post-mortem examination, and the results suggest an autosomal recessive or inherited condition, professionals should ensure that appropriate referrals are made.

## Additional information to consider

***Pre-conception care***

- In general, it is unacceptable to discourage people from becoming pregnant but pre-conception genetic counselling might prepare people for choices
- Carrier tests are available for couples with a previously affected child. However, in order for this to be an option, there must be stored DNA from a previously affected child and genetic

testing should have identified the gene cause in the family. Therefore, it is essential to store DNA from any previously affected children as without this genetic testing cannot be performed. Pre-natal diagnosis cannot be offered.

- Pre-implantation genetic diagnosis - i.e. selection of unaffected fetus. This is available for a number of conditions however, there are criteria that families have to meet before this can be offered. Without a genetic diagnosis of a previous child this cannot be offered, again underlining the importance of stored DNA.
- Pre-conception genetic counselling should be available to help families make the choice that is right for them and to enable them to understand their risks of a future pregnancy being affected.
- Where a deleterious gene has been identified, there is value in knowing whether this couple were aware of the deleterious gene in the family or not; and, if not, whether there were relatives already known to the genetics service with this condition and whether therefore there might have been a 'missed opportunity' to share this information with this couple and potentially offer them carrier testing prior to conception. Genetic information is important not only for this couple, but for other couples in the family as this will help them to make decisions before they have a first affected child.
- The chance of getting a genetic diagnosis is now very good and results can be available within 2 weeks if requested urgently (for example, if a woman is pregnant and requests pre-natal diagnosis) once the samples are obtained from the affected child and both parents.

***Antenatal diagnosis and making decisions to continue with the birth of an affected child:***

- Selective termination is acceptable in the English system, but it is only an option if a test is available to confirm (high probability) the pregnancy is affected.
- Here it will be of value to know whether there was a test available for the condition, and, if this was a second affected child, whether DNA was stored and made available for genetic testing.
- Here it will also be of value to know whether the couple had access to the moral and religious guidance they needed to support their decision to continue with or terminate the pregnancy; it will be useful to know which agency counselled them, whether there was culturally competent, sensitive clinical input alongside this and whether they felt they had all the information about the prognosis of the child to make their decision.

***Post-birth:***

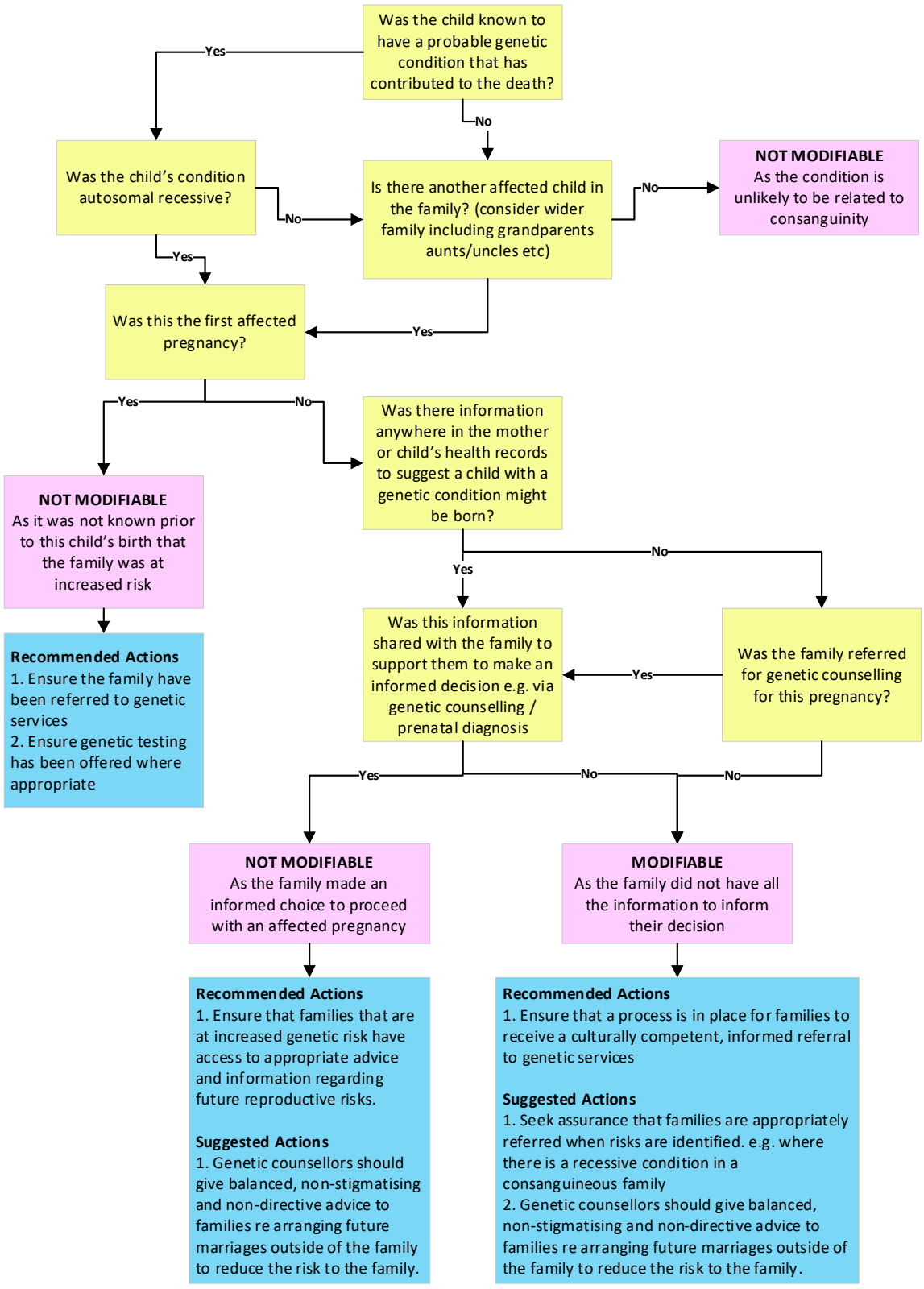
- Action to avoid infant death is clearly acceptable but there are limited effective options for the serious conditions. And this may simply push death back a few months/years (so may not be an infant death but a child death) and may need to consider the consequences of longer-term care of a severely disabled child for family and healthcare costs. This again underlines the importance of stored DNA for genetic testing (see section on pre-conception care).
- If pre-birth testing is available, it may give some advantage in acting early once a child is born, but again, the options are currently limited
- Here it will be important to know about the child's care, and whether there was any delay in treatments and for what reason; as above, knowing whether there was information in the multi-agency system about the potential for an affected child to be born would be helpful;

information about whether parents were counselled about potential birth and treatment options etc.

- If the taking of DNA samples is being offered after the child has died it is important that parents know that taking DNA samples is not as invasive as the post-mortem process and can be done by taking a blood sample or small piece of skin. It is also important that the family understand that this is not being done for research purposes, but to assist with decisions for any future pregnancy, as this may impact their decision to give consent.

## Appendix A: Decision making tool for determining modifiability relating to consanguinity in CDRM or CDOP review.

**Decision making tool for determining modifiability relating to consanguinity in CDRM or CDOP review**



## Appendix B: References


1. Sheridan, E. et al (2013) “Risk factors for congenital anomaly in a multi-ethnic birth cohort: an analysis of the Born in Bradford study” The Lancet, Vol 382, Issue 9901, p.1350 – 1359, October 19, 2013
2. Teeuw, M.E. et al (2010) “Do consanguineous parents of a child affected by an autosomal recessive disease have more DNA identical-by-descent than similarly related parents with healthy offspring? – Design of a case control study” BMC. Med. Genet 2010 Jul 16; 11:113