Supporting Material: Understanding consanguinity-related child deaths

1. Methodology and limitations

Cohort identification

All children (0 – 17 years inclusive) reported to the National Child Mortality Database (NCMD) who died between 1 April 2019 and 31 March 2023, where the death had been reviewed and finalised by a Child Death Overview Panel (CDOP) before 27 November 2025, were included for analysis.

Deaths of children born to consanguineous parents were identified if:

i. consanguinity was recorded as a factor on the analysis form completed by the CDOP. This included factors graded as 'unlikely to have contributed to the death' and also included those that 'may have contributed to vulnerability, ill health or death', or

ii. the reporting form question 'Are the child's parents related to each other (E.g. Cousin)?' was answered yes, and there was other information in the case record to confirm consanguinity status.

These steps were taken to complete identification and appropriate inclusion of all cases, ensuring that the cases included for analysis had either consanguinity recorded by the CDOP or other information existed in the record to confirm consanguinity status. This ensured that cases included in the 'consanguinity' group were accurately reported.

Deaths that did not meet the criteria outlined above were included in the 'non-consanguineous' group in the analyses, and this group included deaths where consanguinity was reported as 'not known'.

The methodology outlined here was developed for this thematic report and therefore numbers reported may differ to other NCMD outputs.

Data sources

The NCMD collects data on all children in England who are born alive (at any gestational age) and die before their 18th birthday, excluding stillbirths and legal terminations of pregnancy. For more information, please see our website.

Hospital Episode Statistics (HES) admitted patient care data were used to identify children that had been diagnosed with a life-limiting condition. The methodology of identifying these conditions is available in the supporting material for the NCMD Thematic Report on Children with Life-Limiting Conditions and Palliative and End-of-Life Care Needs. ICD-10 coded diagnoses available in HES data may not capture rare genetic diseases. However,

this work only used HES data to identify life-limiting conditions, and HES data was not used to identify the cohort of deaths due to chromosomal, genetic and congenital anomalies, which was identified using the category of death recorded by CDOPs. Please note that the HES data supplied from NHS England was under a new agreement and methodology, therefore data differs slightly to that reported for the NCMD Thematic Report on Children with Life-Limiting Conditions and Palliative and End-of-Life Care Needs. This is due to improved data linkage rates.

For deprivation measures, the child's postcode of residence was linked to the Index of Multiple Deprivation, with a lower value suggesting greater deprivation.

Other limitations

The report describes the number and characteristics of deaths of children who were born to consanguineous parents in the context of all child deaths. As there is no national population data on all children born to consanguineous parents, the risk of child death is not reported, and the analysis is a description of the deaths that occurred only. Further, the report includes only children born to consanguineous parents who died before their 18th birthday, and does not include stillbirths or lawful terminations of pregnancy, as these are not subject to the child death review process. It is also important to recognise that any patterns observed in the data may reflect a complex interplay between consanguinity and other determinants of health, including maternal, socioeconomic, cultural, or health system factors, all of which may influence child mortality.

The NCMD is reliant on accurate data being inputted by professionals involved in child death reviews. This includes ethnicity of the child, the category of death recorded by CDOPs (used to identify the cohort presented in sections 2 and 3) and the presence of consanguinity. All deaths where the parents were reported as consanguineous and confirmed using the methodology outlined were included. 'Consanguinity' is not closely defined in the child death review data collection forms, and therefore all different degrees of relationships are assumed to be included (e.g., first cousin/second cousin/other) and no specific relationship types were excluded. In addition, there is potential for reporting biases in the data collection around consanguinity in child death reviews which should be considered in the interpretation of data. Consanguinity may be more likely to be investigated and recorded when a child dies from chromosomal, genetic, or congenital anomaly than from other causes of death (e.g., traumatic death), or may be considered more frequently in some societal groups or areas, than others.

Section 1 reports child deaths that occurred between 1 April 2019 – 31 March 2023 (4 years), while sections 2 and 3 report deaths that occurred between 1 April 2019 – 31 March 2022 (3 years). Sections 2 and 3 reported a 3-year time period to ensure clinical coding of genetic conditions was fully completed for this report. Deaths included in all sections of this report were restricted to those that had been reviewed by a CDOP. For deaths that occurred between 1 April 2019 – 31 March 2023, 95% had been reviewed by a CDOP before 27 November 2025. The cohort was restricted to deaths that occurred in this

period because most (95%) of the deaths during this period had been reviewed by CDOP. Therefore, there will be a small underestimation of the number of deaths of children born to consanguineous and non-consanguineous parents in this report, and the total number of deaths may differ to other NCMD reports. As consanguinity is collected within the child death reporting form and analysis form, we were reliant on the information from the completed review to identify this cohort of deaths, so we restricted the time period so that the number of deaths born to consanguineous parents was as complete as possible, and the most complete information was reported.

In 2023, the NCMD convened a Task & Finish group composed of experts in a range of fields from around the country, and worked with them to provide comprehensive guidance for child death review professionals to use when reviewing deaths of children born to consanguineous parents. The majority (82%, n=362/440) of the deaths in section 3 were reviewed before the publication of this guidance (January 2023).

Category of death

CDOPs are required to assign a category to each death during the review. Information on this categorisation process can be found in the child death analysis form. While more than one category can be applied, the primary category of death is the uppermost category of death selected. Sections 2 and 3 of this report provide analysis of the deaths where *Chromosomal, genetic and congenital anomalies* was selected as **any** category of death by the CDOP (i.e., not only deaths where this was recorded as the primary category).

Coding of clinical conditions and recurrence

Data linkage with the National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) could not be achieved within the timeframe for this report, therefore NCMD undertook clinical coding of genetic conditions. This was undertaken by two clinicians with relevant expertise, for the purposes of this thematic report. Information recorded in the data fields: suspected cause of death, notification details, circumstances of death, medical certificate cause of death, and cause of death as recorded by CDOP, was used to assign a category to the clinical condition. The categories used (as presented in Table 7) were mutually exclusive.

The category *congenital anomalies* included deaths where the baby was born with an anomaly and no genetic testing had been done to confirm a genetic aetiology, or there may not be suitable testing that can be done (e.g., diaphragmatic hernia). Each death was coded in the most suitable category. Many babies had multiple congenital anomalies but also had an overarching unifying genetic aetiology, and therefore the case was coded under the appropriate category (e.g., autosomal dominant disorders). The category *not genetic, congenital or insufficient information* was used if upon assessing all available information there was no evidence for a genetic aetiology (e.g., pneumonia), and another category was not appropriate, or if there was insufficient information available to categorise the case.

Information within the NCMD record was also searched for evidence of recurrence (i.e., whether there was a history of this condition in the family), and where no information could be identified no recurrence was assumed. Therefore, recurrence will potentially be underestimated.

2. References

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