NCMD

National Child Mortality Database

Knowledge, understanding and learning to improve young lives

Consanguinity in CDRMs and CDOP reviews

Wednesday 1 February 2023

10.00am to 11.15am

Presentation will start at 10.05 to allow participants time to join

Consanguinity in CDRMs and CDOP reviews

Vicky Sleap
NCMD Programme Manager

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National Child Mortality Database

Knowledge, understanding and learning to improve young lives

What will this webinar cover?

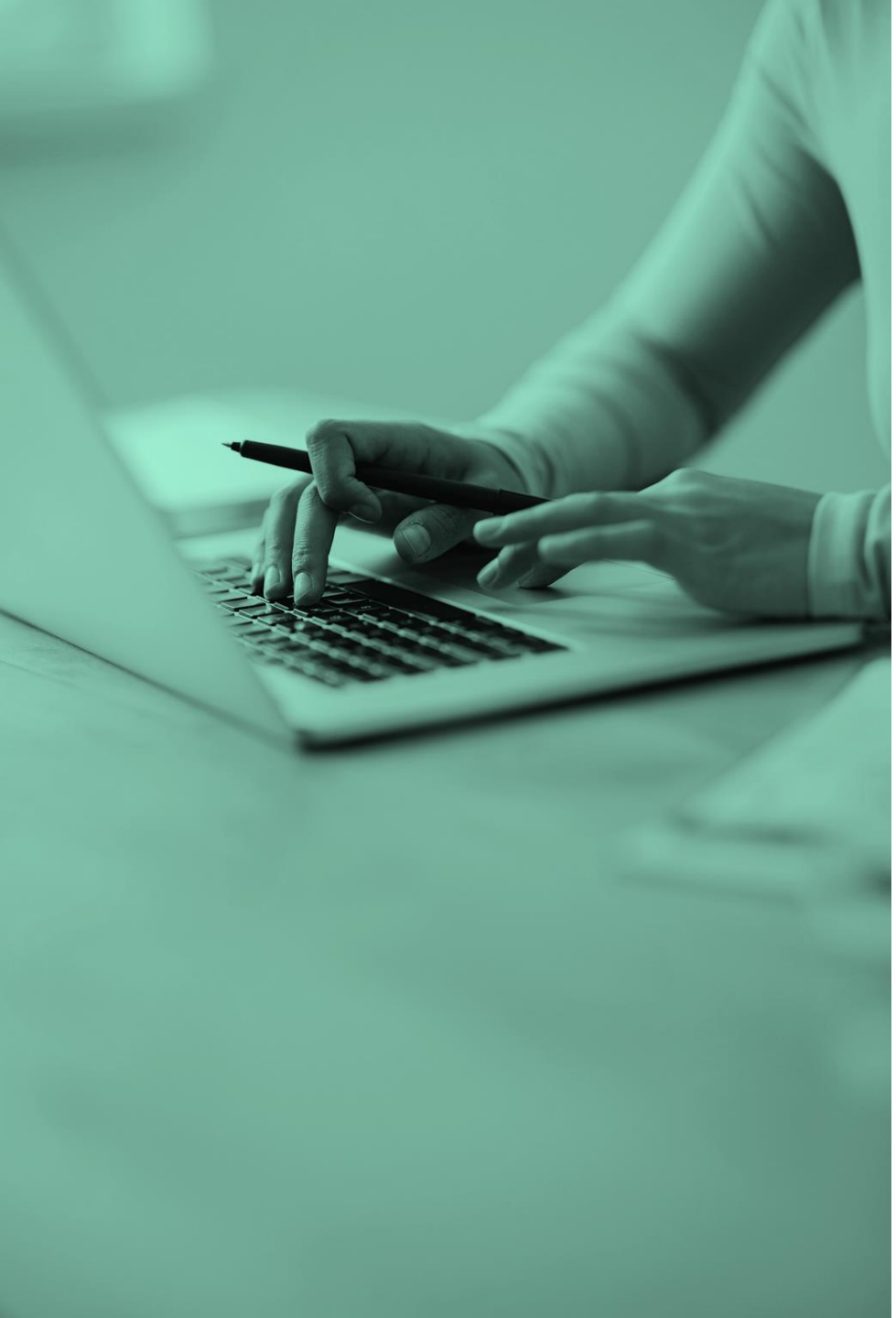
- Background to this issue and how the national group was convened
- Presentation from Naz Khan, Clinical Lead Equality, Ethnicity and Genetics at NHS England and NHS Improvement
- NCMD guidance for CDR professionals on consanguinity and
- National strategy on consanguinity and support for families

Acknowledgements

Nasaim Khan (Chair of the Task & Finish Group)	Clinical Lead Equality, Ethnicity & Genetics, Maternity Transformation Programme, NHS England & NHS Improvement
Christine Brown	Deputy Director of Quality & Safeguarding, NHS England North East & Yorkshire
Louise Clarkson	SUDIC / CDOP Manager, Bradford
Stephanie Davern	Project Manager, Manchester City Council, Manchester Population Health
Daniel Devitt	Senior Public Health Strategist, West Berkshire Public Health, Reading Borough Council
Joanna Garstang	Designated Doctor for Child Deaths, Birmingham
Noreen Gurner-Smith	Designated Nurse for Child Death Reviews, Surrey Child Death Review Partnership
Rezvana Hassan	Lay Representation
Catherine Kearney	Designated Doctor for Child Deaths, Luton
Sheila Moore	Chair of North & South of Tyne CDOP
Eduardo Moya	Consultant Paediatrician, Bradford Teaching Hospitals NHS Foundation Trust
Denise Phillip	Designated Nurse for Safeguarding Children, Kirklees Health & Care Partnership
Vicki Stadnicki	Public Health Manager, Kirklees Council
Sarah Steel	Designated Doctor for Child Deaths, Norfolk & Waveney Integrated Care Board

Background

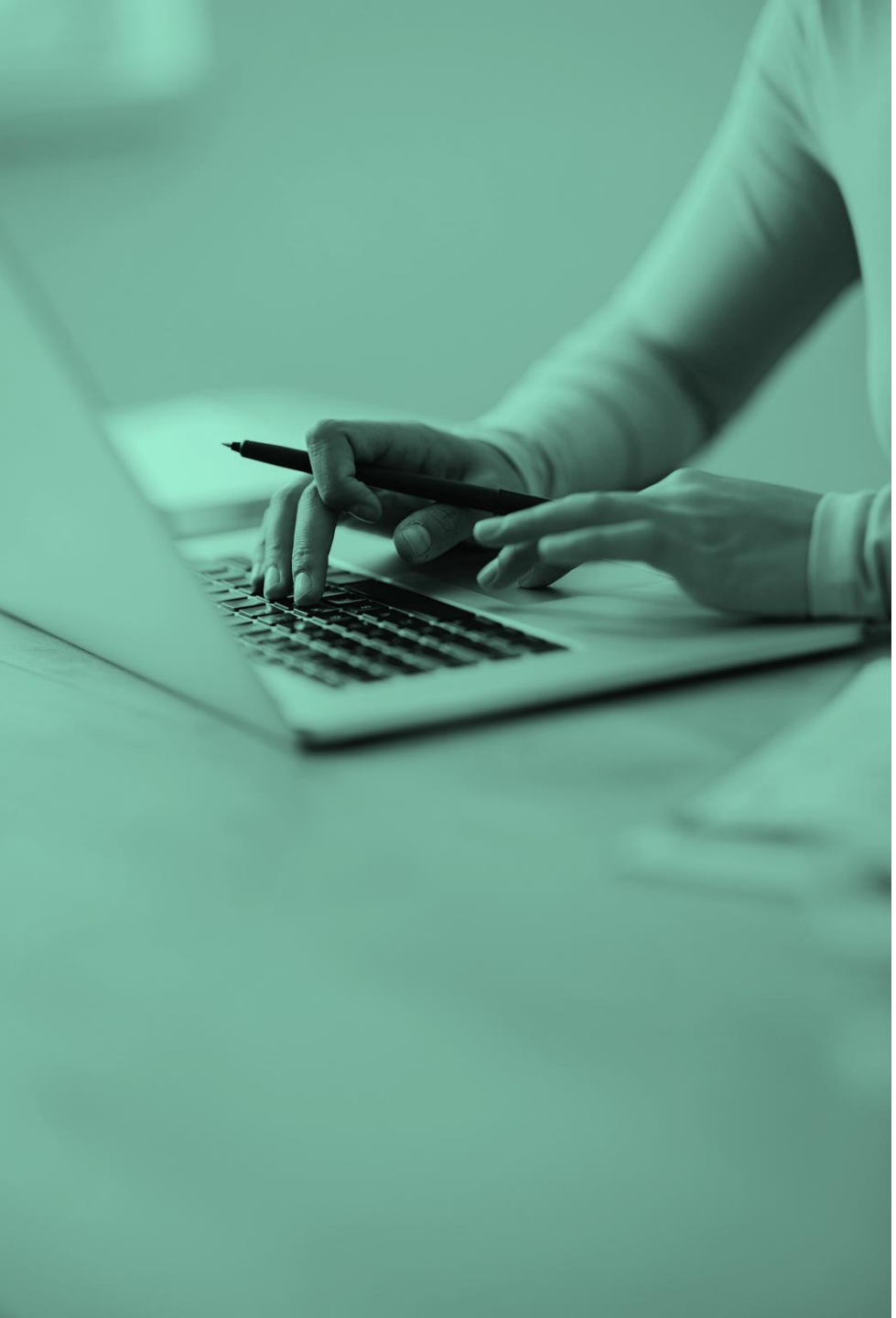
- How to assess consanguinity, also known as **close relative marriage,** is the most common question NCMD receives from CDR professionals
- There is a strong desire from CDR professionals to balance the need to be consistent and culturally sensitive with the need to identify and implement learning
- The NCMD Second Annual Report highlighted that consanguinity was one of the most frequently recorded modifiable factors by CDOPs, however in most cases there was no detail provided about how this decision had been made or what interventions were being considered in the local area



National Steering Group on Consanguinity & Genetic Risk

- Following the publication of the report, NCMD was contacted by the then Chair of the National Steering Group on Consanguinity and Genetic Risk who challenged the quality of CDOP data in this area
- Concerns were:

- the variability with which consanguinity was recorded and graded
- in what circumstances it was considered modifiable
- An apparent lack of understanding of consanguinity and the worry that publishing data in this context would contribute to increased stigma for families



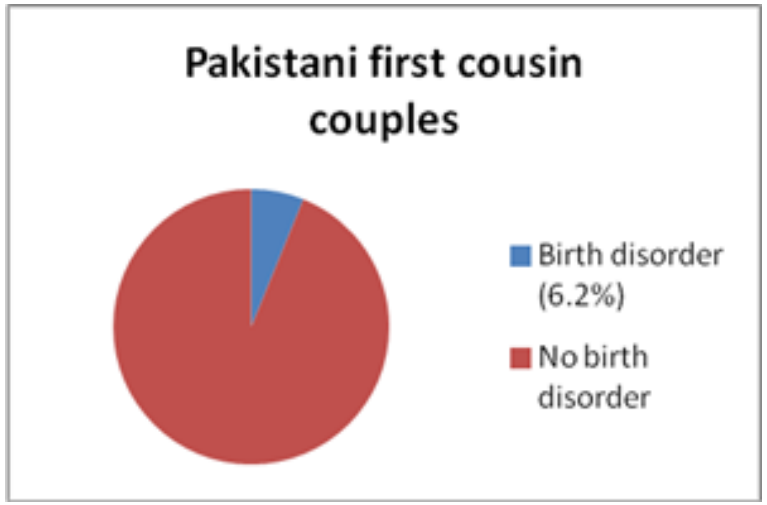
CDOP concerns

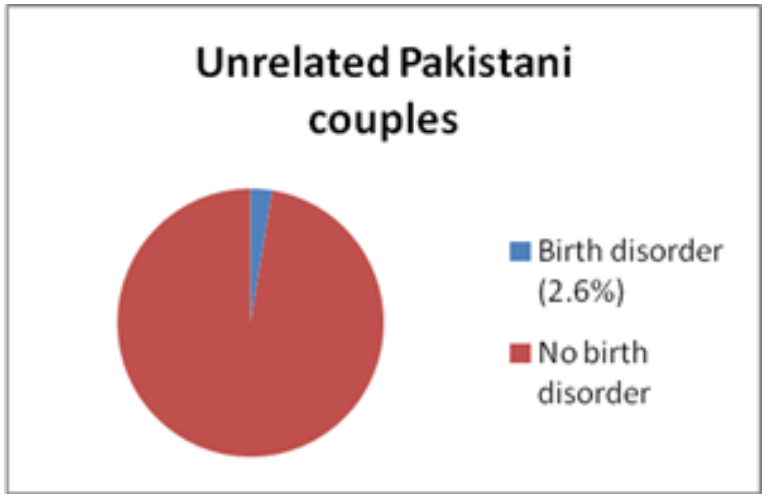
- At the same time as this was happening, NCMD was receiving a number of communications from CDOPs across the country expressing similar concerns
- Colleagues in CDOPs in the North East and Yorkshire held a learning event where this issue was discussed and were keen to set up a Task & Finish Group to develop some guidance in this area
- The group was set up with representation from a wide number of CDOPs, NHS England, NHS Improvement, the National Steering Group on Consanguinity and Genetic Risk and lay membership representing families with lived experience of the issue.

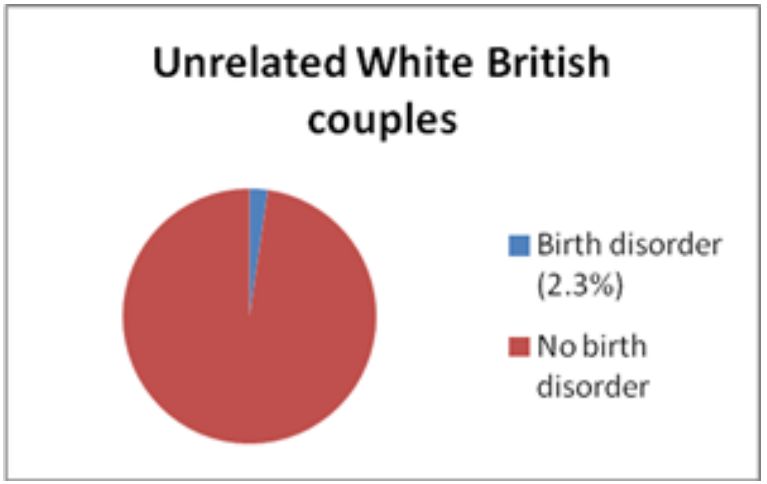
Naz Khan,
Clinical Lead Equality, Ethnicity and Genetics at NHS England and NHS Improvement

Risks of genetic disorders in different populations

- Risks of infant death & disability higher among communities practising close relative marriage.
- All Asian British/Pakistani heritage children were over-represented in both mortality and chronic morbidity categories (MBRRACE-UK 2020, PHE, CDOP)
- Congenital abnormalities are the leading cause of death for Pakistani infants (Li et al 2018)
- Family-level clustering
- Over 90% of babies born to cousin couples are healthy.







Sheridan et al 2013; The Lancet

Close Relative Marriage

- Consanguineous marriage = marriage between blood relatives
- Benefits and risks recognized for centuries
- Close relative marriage is widely practised globally with recognized benefits to couples and their families.
- 20% of world's population live in communities that favour consanguineous marriage
- 8.5% of all births are to parents who are consanguineous
- Associated with Islam but neither encouraged nor discouraged by Islam
- However, the level of increased risk has often been exaggerated and this marriage pattern has been stigmatised in the UK

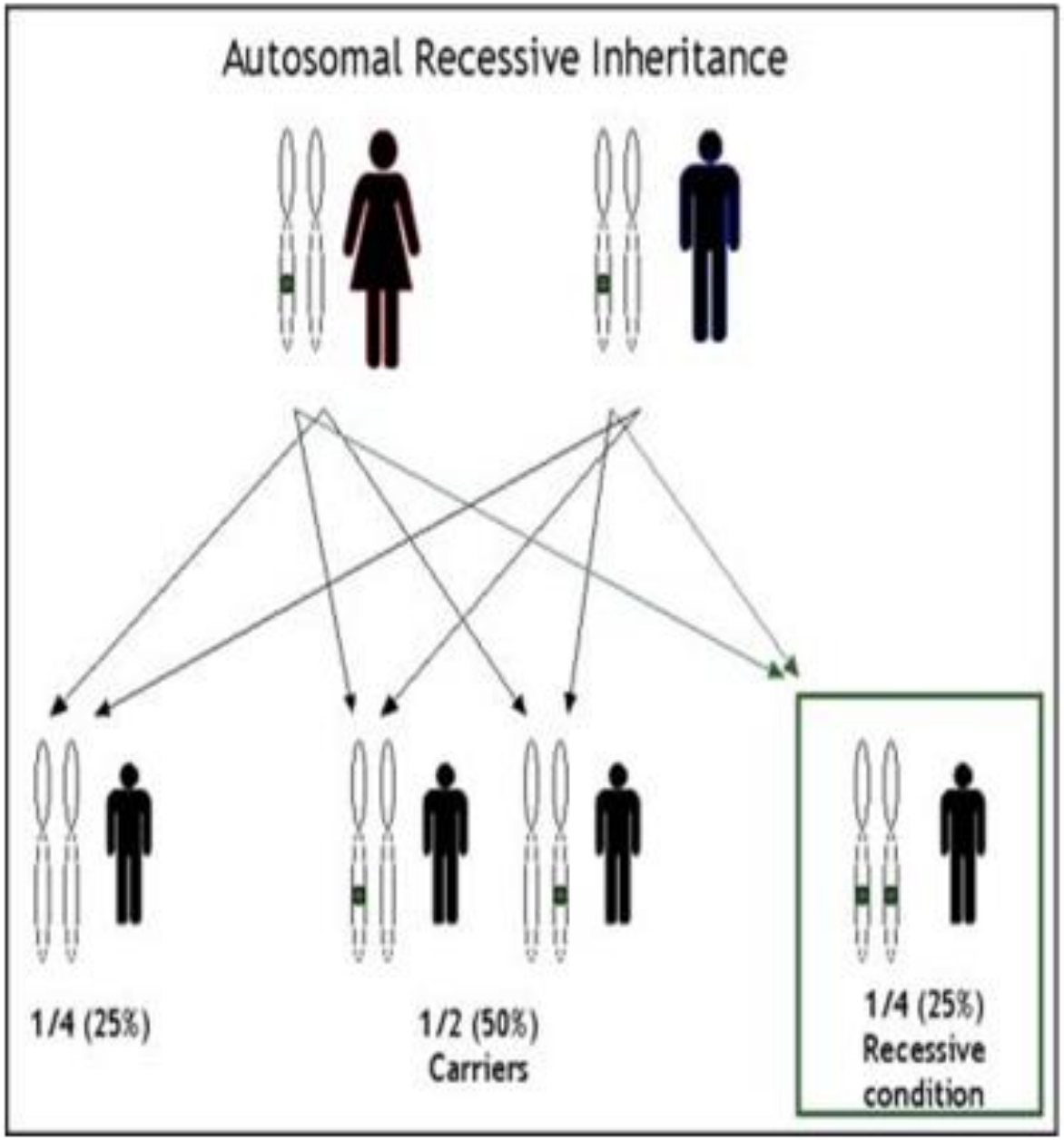
UK Patterns of Close Relative Marriage

- Born In Bradford study (2007-2011)
  - 12,453 women
  - 50% identified as South Asian
  - 49.9% non South Asian
- Pakistani mothers
  - 37% married to a first cousin,
  - 21% to other blood relative and
  - 42% to non-relative
- Also common in other ethnic groups, but lack of data

Genetics

- An autosomal ***recessive*** genetic condition occurs when both copies of the SAME gene happen to be faulty
- Recessive genetic conditions occur in all population groups
- Some recessive conditions are quite common
- Partners in a consanguineous relationship are not more likely than other individuals to carry faults in their genes
- But, because they share more of their genetic material than unrelated partners, they are more likely to carry the same faulty gene as their partner
- Children of related partners have a higher risk of inheriting two copies of the same faulty gene for a recessive genetic condition than children of unrelated partners

Genetics


Autosomal Recessive Inheritance
1/4 (25%)
1/2 (50%)
Carriers
1/4 (25%)
Recessive
condition

Pakistani Population

Data from: research; Child Death Overview Panels; and clinical experience (midwives, genetics, health visitors and social care); Audit; PPI

- Repeated unexpected affected births (and deaths) to couples and across extended families
- Significant number attributed to recurrence in same family
- More than 50% of families with a likely AR condition are not referred to Genomic service
- Can lack the confidence to seek services and rely on referrals by healthcare practitioners
- Are sometimes refused referrals by GPs and others
- Have mixed experiences of genomic services, with some leaving without a good grasp of information & choices
- May struggle to share information with family members, but can be supported to do so
- Persistent unmet need for information and support
- Positive outcomes where services are better

Purpose of the NCMD 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 ensure enhanced equity of access to appropriate, culturally competent, clinical genetic services for all families at increased genetic risk, to enable informed decision-making.

Information Collection

- 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.
- NCMD is working on updates to the supplementary form based on recommendations from the Task & Finish Group

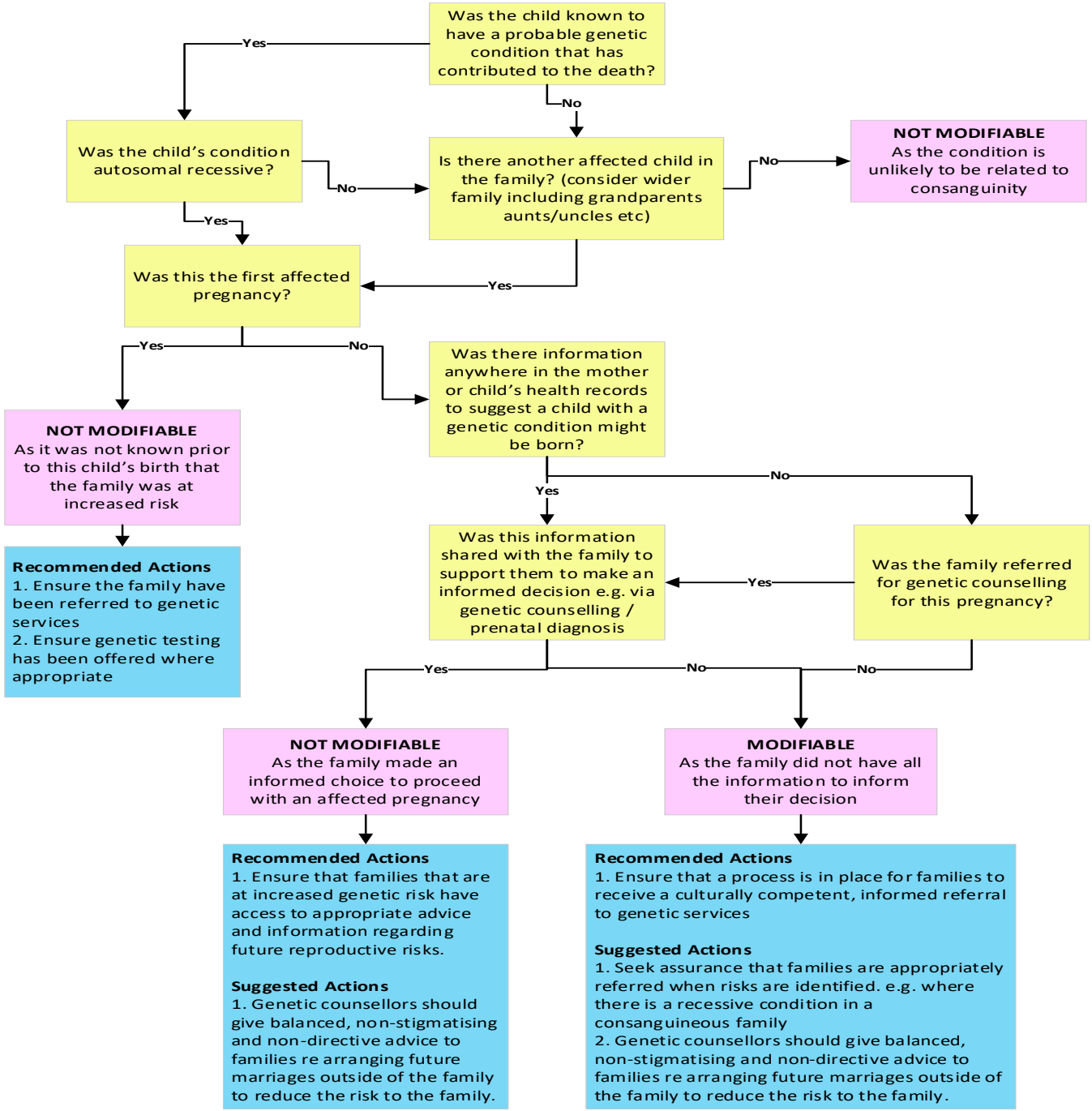
Consanguinity as a contributory factor

- Consanguinity can be contributory but not 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. This will primarily be through access to genetic counselling services
- 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.
- It is important to consider whether the family had all the information they needed to make an informed choice about their pregnancy.

Consanguinity as a modifiable factor

- Consanguinity itself should not be considered a modifiable factor. Instead you 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.

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



Naz Khan,
National Strategy on Consanguinity

National Work

- To reduce unmet need for genetic counselling and testing
- To increase informed reproductive decision making (within existing unions and future unions)
- Strengthen access and ensure cultural competence
- Identify families with an affected member
- Cascade information and support wider family members

9 Areas of High need

Culturally Competent Genetic Services: 4 Strands

- Community-led
- Raise awareness of genetic risk
- Improve access to services
- Empower and build trust
- Varied, accessible format
- Reduce stigma and fear
- Local, multi-agency working group established
- *Nationally-provided health promotion materials*

1. Raise genetic literacy at community level

2. Educate & equip healthcare professionals

- Close relative marriage midwife "champion" 0.4WTE band 6
- Added value - health visitor champion (not required)
- Promote the e-learning module
- *e-learning for healthcare 'close relative marriage' modules*
- *National webinar post training*

All four strands must be applied to gain best results

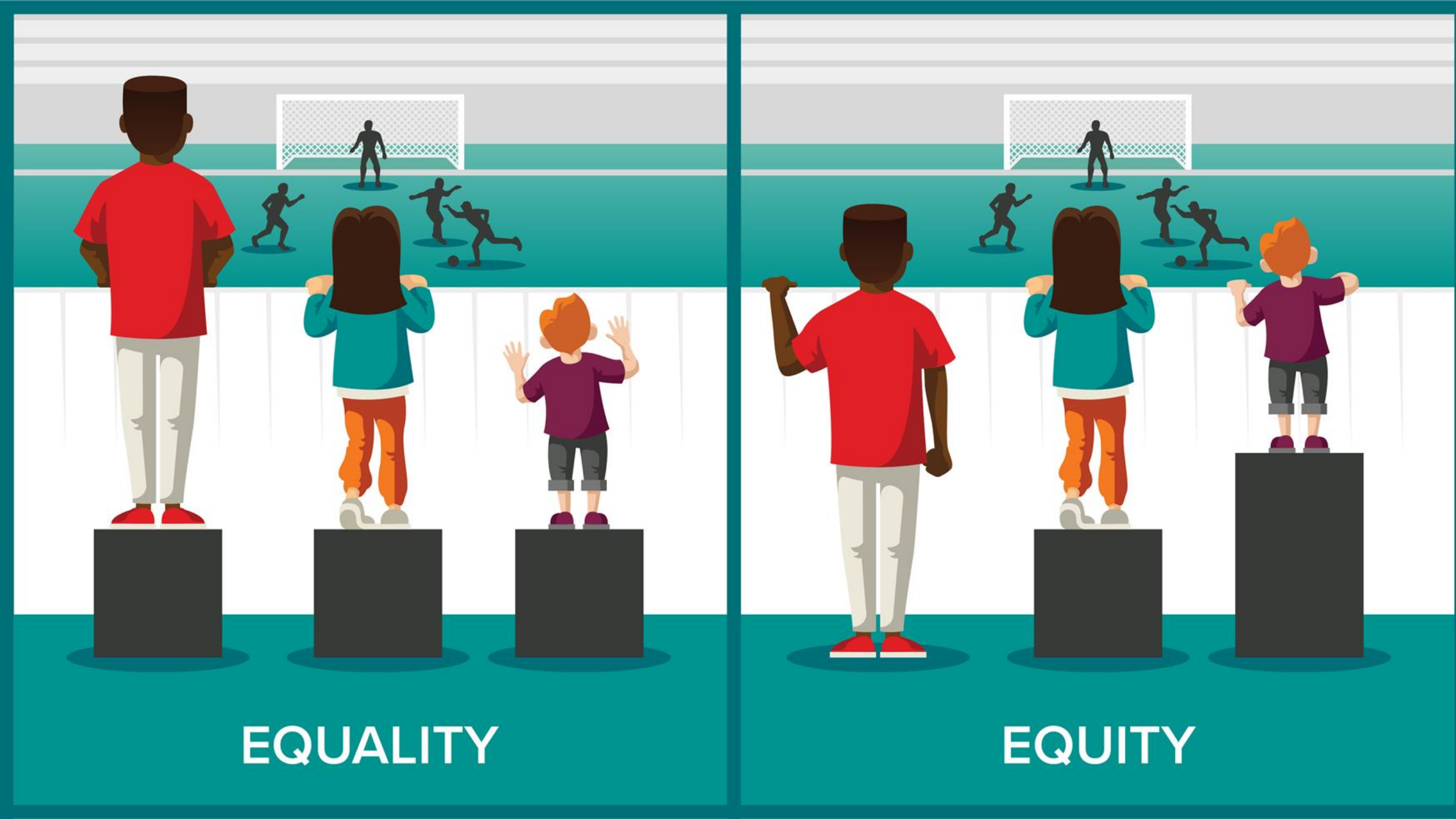
- Develop and improve care pathways from primary & secondary care – improve access for underserved groups
- *Genomics associates (nationally funded) in clinical genetics services to support access, follow up non-attenders & support family conversations*

3. Improve access to genetics services for underserved groups

4. National support

- Oversight: national steering group
- National communities of practice for close relative marriage midwives, genomics associates & public health teams
- Progress reports & share learning
- Metrics with analytics advice
- Evaluation
- Funding: ongoing & start up

Key:
National support offer


EQUALITY
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Questions

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