

Contribution of congenital disorders to under-5 mortality

To the Editor: The article ‘Where do children die and what are the causes?’, which appeared in the April 2016 issue of the *SAMJ*,^[1] provides an overview of the causes of death in under-5 children in the Metro West geographical service area of the Western Cape for 2011. It highlights the proportion of under-5 deaths from congenital abnormalities (obvious structural abnormalities), which are particularly prevalent in early neonatal mortality – a close third (9.6%) of in-hospital deaths after hypoxia (10.0%) and immaturity (40.6%) according to the Perinatal Problem Identification Programme (PIIP) data.^[1] In the Local Mortality Surveillance System in-hospital data, congenital abnormalities are ranked as the second (13.5%) cause of early neonatal death after prematurity (35.8%).^[1]

Although already prominent as a cause of death, congenital disorders (CDs) may collectively contribute to a greater proportion of child deaths than reported. In the study, congenital abnormalities relate to chapter XVII: Congenital malformations, deformities and chromosomal abnormalities in the *International Classification of Diseases* (ICD-10) and are aggregated to the Burden of Disease list of causes.^[1-3] Limited to developmental structural abnormalities only, this excludes a significant portion of CDs found elsewhere in the ICD-10 system (e.g. congenital syphilis A50; haemophilia D66 - 68; oculocutaneous albinism E70.310).^[3] This ICD-10 coding fragmentation has exacerbated global confusion around terminology related to CDs.^[4] In 2006, international agreement was reached on the synonymous use of the terms CDs and birth defects, defined as abnormalities of structure or function, including metabolism, present from birth and manifesting at birth or later in life.^[5] However, use of inequivalent terms, such as congenital abnormality, continues. Consequently, data for subsets of CDs are often interpreted as the totality of CDs when it is not the case. If single-gene disorders, accounting for 30.0% of CDs (B Modell – personal communication, 2016), were pooled with congenital abnormalities in the study by Reid *et al.*,^[1] a greater proportion of under-5 deaths may be attributed to CDs.

CDs are also significantly underestimated, as many remain undiagnosed or are misdiagnosed, with the incorrect cause of death due to the lack of trained clinicians,^[6-8] as acknowledged by the Child Healthcare Problem Identification Programme.^[4,9] Undiagnosed CDs may be the underlying cause of death in a number of cases assigned to other causes (including ‘ill-defined’) or comorbidity in the study, as infants born with CDs, such as congenital heart defects, may be more susceptible to infection.

Honein *et al.*^[10] reported that CDs are more than five times as likely to occur among very preterm infants (24 - 31 weeks) compared with

term infants (37 - 41 weeks), resulting in 16.0% of preterm infants having a CD. A significant portion of deaths assigned to ‘prematurity’ in early neonatal deaths in the study may therefore be undiagnosed CDs, which are predisposed to preterm birth.

The relative frequency of CDs should be noted and investigated in light of the abovementioned factors contributing to their under-reporting, particularly as the majority of CD-related deaths occur during the first 5 years of life. As the proportion of deaths from CDs increases along with epidemiological transition – seen in the dramatic decrease in HIV-related deaths – the challenge of CDs will continue.^[11] Possible areas for further study include comparison with other provincial populations, analyses of preventable CDs (e.g. fetal alcohol spectrum disorder) and prenatal diagnosis of serious CDs.

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