Contribution of congenital disorders to neonatal mortality in South Africa

To the Editor: The article 'Reducing neonatal deaths in South Africa: Progress and challenges'[1] in the March 2018 *SAMJ* Maternal and Child Health Supplement reviews recent neonatal estimates, causes and ways of reducing preventable deaths in this age category in the context of the Sustainable Development Goals.[2] While the article acknowledges the increasing contribution of 'congenital anomalies' as a cause of neonatal death, it fails to acknowledge the proportion of 'congenital disorders' (CDs) as a collective. No definition is included, but the literature defines congenital anomalies as macroscopic morphological anomalies present at birth,[3,4] referring to clinically obvious structural abnormalities as classified in Chapter XVII: 'Congenital malformations, deformations and chromosomal abnormalities' of the *International Statistical Classification of Diseases and Related Health Problems* (ICD-10).[5] This excludes 'non-syndromic congenital disability (intellectual, physical, visual and auditory disability and epilepsy), common single gene disorders such as the haemoglobin disorders, glucose-6 phosphate dehydrogenase deficiency, cystic fibrosis, oculocutaneous albinism, spinal muscular atrophy and inborn errors of metabolism'[4] and teratogens. These are distributed throughout the ICD-10 system, accounting for a third of CDs globally.[6] Presenting only a subset of CDs to represent the totality of CDs has implications, particularly for cause-of-death rankings and planning of services and interventions.

While the role of congenital anomalies is indicated as a notable cause of death in neonates in South Africa (SA), ranking fourth after prematurity, intrapartum-related events and infection, CDs may rank higher as a cause of death. CDs are internationally defined as abnormalities in structure or function present from birth, including inborn errors in metabolism.[4] This classification disparity is further evidenced by the average baseline birth prevalence rates of congenital anomalies of 20 per 1 000 live births[7] v. that of CDs at >39.7 per 1 000 live births.[6,8] Interestingly, contrary to the article text, data in Fig. 5 showing causes of neonatal deaths per level of care indicate that congenital anomalies rank third as a cause of neonatal death at an overall 8.8%, ahead of infection at 7.7%, with the greatest disparity between these two causes of death at district hospitals.[1]

Non-diagnosis or misdiagnosis resulting in the cause of death being incorrectly attributed also contributes to underreporting of CDs as a cause of death. Honein *et al.*[9] report CDs as being twice as common in preterm infants (24 - 36 weeks) as in term infants, and five times more likely in very preterm infants (24 - 31 weeks), with 16% of very preterm births having a CD. Similarly, a portion of deaths assigned to intrapartum-related events may be due to severe congenital cerebral palsy (CP). The Centers for Disease Control and Prevention indicates that 85 - 90% of CP is congenital,[10] of which an estimated 30 - 40% has a genetic aetiology, with birth asphyxia only accounting for 10 - 15% of all CP cases.[11-16] Congenital syphilis, a CD by definition, contributes a minimum of 3% of neonatal deaths[17] and together with other congenital (TORCH) infections (which include toxoplasmosis, other (syphilis, varicella-zoster, parvovirus b19), rubella, cytomegalovirus and herpes) may also be allocated to the infections cause of death category.[8] This proportion is probably an underestimate, since diagnosis of congenital syphilis in neonates is more difficult than in pregnant women, with adverse pregnancy outcomes in 38% of cases of maternal infection, including neonatal death in 7% of cases.[18]

Neonates born with a CD have an increased susceptibility to infection in certain instances, with the infection being more 'obvious' and more likely to be diagnosed and assigned as the cause of death than the underlying CD in cases of comorbidity.[19,20] The lack of a newborn screening policy and top-to-toe examination of the infant prior to discharge further contributes to non-diagnosis and the inaccurate assessment of the contribution of CDs to the burden of disease.[8,19,21,22]

With a significant proportion of under-5 deaths occurring during the neonatal period, comprehensively addressing CDs as a cause of neonatal deaths in SA is now an imperative. World Health Assembly (WHA) Resolution 63.17 of 2010[23] recognised the importance of CDs as a cause of stillbirths and neonatal mortality and their contribution to the failure in attaining Millennium Development Goal 4. The same WHA recommendations must now be heeded if the SGD targets set for 2030 to end preventable deaths in this age group are to be met.[2,23]

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