Understanding the burden of congenital disorders in South Africa: data, trends, and implications

NDOH World Birth Defects Day Webinar

3 March 2026

Extraordinary Associate Professor Helen Malherbe
Centre for Human Metabolomics
Desmond Tutu School of Medicine
Faculty of Health Sciences, North-West University
Potchefstroom, South Africa

Disclosure

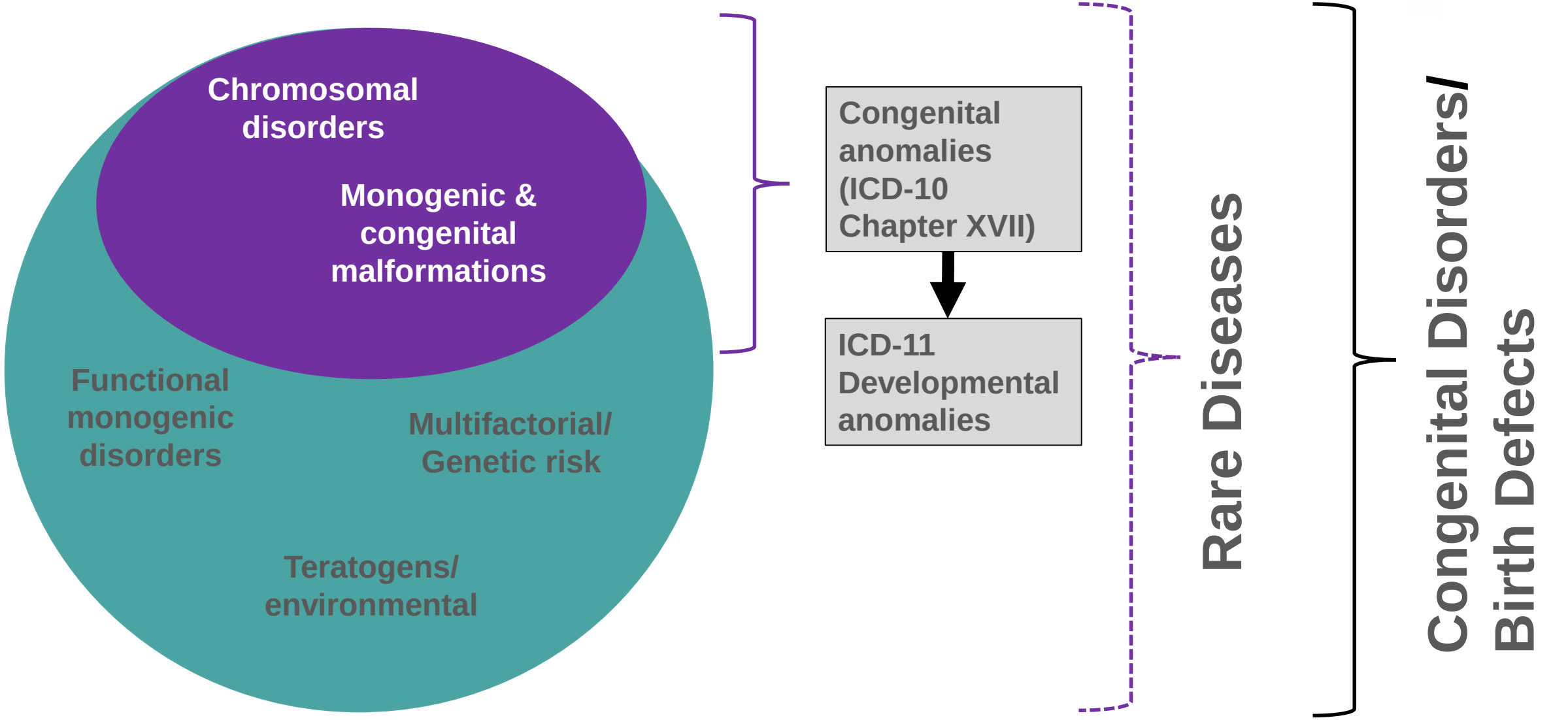
- Employee of North-West University;
- Speaker for Sanofi, Astra Zeneca (2025/6);
- Consultant for IPASA (2024/5);
- Commissioner: Lancet Commission on Rare Diseases;
- Vice Chair, Inter-disciplinary Scientific Committee, International Rare Diseases Research Consortium (IRDiRC);
- Member of Rare Disease Access Initiative (RDAI);
- Member of Undiagnosed Diseases Network International (UNDI).

Madeleine Eva Malherbe
14 October – 24 December 2004

Contents

1. Definitions: what are congenital disorders?
2. Why do CDs matter?
3. Global context and trends
4. South African data context
5. Epidemiological context – why CDs matter now
6. Economic & health system impact
7. Current service landscape
8. Implications – what should we focus on?
9. Global momentum
10. Concluding remarks

Definition: What are congenital disorders?



Congenital disorders = Birth defects ≠ Congenital anomalies (WHO, 2006)

Definition: Congenital Disorders & Rare Diseases

World Health Organization

Birth Defects = Congenital disorders:

"abnormalities in structure or function (including metabolic disorders) present at birth" (WHO 2006)

- A heterogeneous group of structural, functional, genetic, metabolic and congenital infection – related conditions.
- Occur in an estimated **3–6% of all births -** individually diverse, but collectively a major public health concern.
- All are present at birth but only some are visible/detected/diagnosed then; others (e.g., metabolic, cardiac, developmental conditions) may only manifest later.
- A leading contributor to **neonatal mortality, under-5 mortality, and lifelong disability**.
- The myth – "**nothing can be done…"**
- But: **70% of birth defects can be prevented, effectively cured & disability mitigated** through optimal **[community] genetic services** (Cziezel et al, 1995).

Why do CDs matter?

- A major contributor to neonatal and child mortality & lifelong disability;
- Present at birth – affecting the most vulnerable of society;
- Increasing relative importance as infectious causes of mortality decline;
- Many are preventable, treatable or mitigated, through:

Primary prevention:
Before pregnancy

Secondary prevention:
During pregnancy

Tertiary prevention/care:
From birth

- They require system wide response (not solely specialist care), including:
  - Primary heath care (ANC, family planning, immunisation etc);
  - Maternal and newborn/child services;
  - (Paediatric) surgery capacity;
  - Rehabilitative care (feeding-, speech- physio- therapy etc);
  - Palliative & end of life care;

Addressing CDs is central to achieving Sustainable Development Goal 3.2 targets (reducing neonatal mortality).

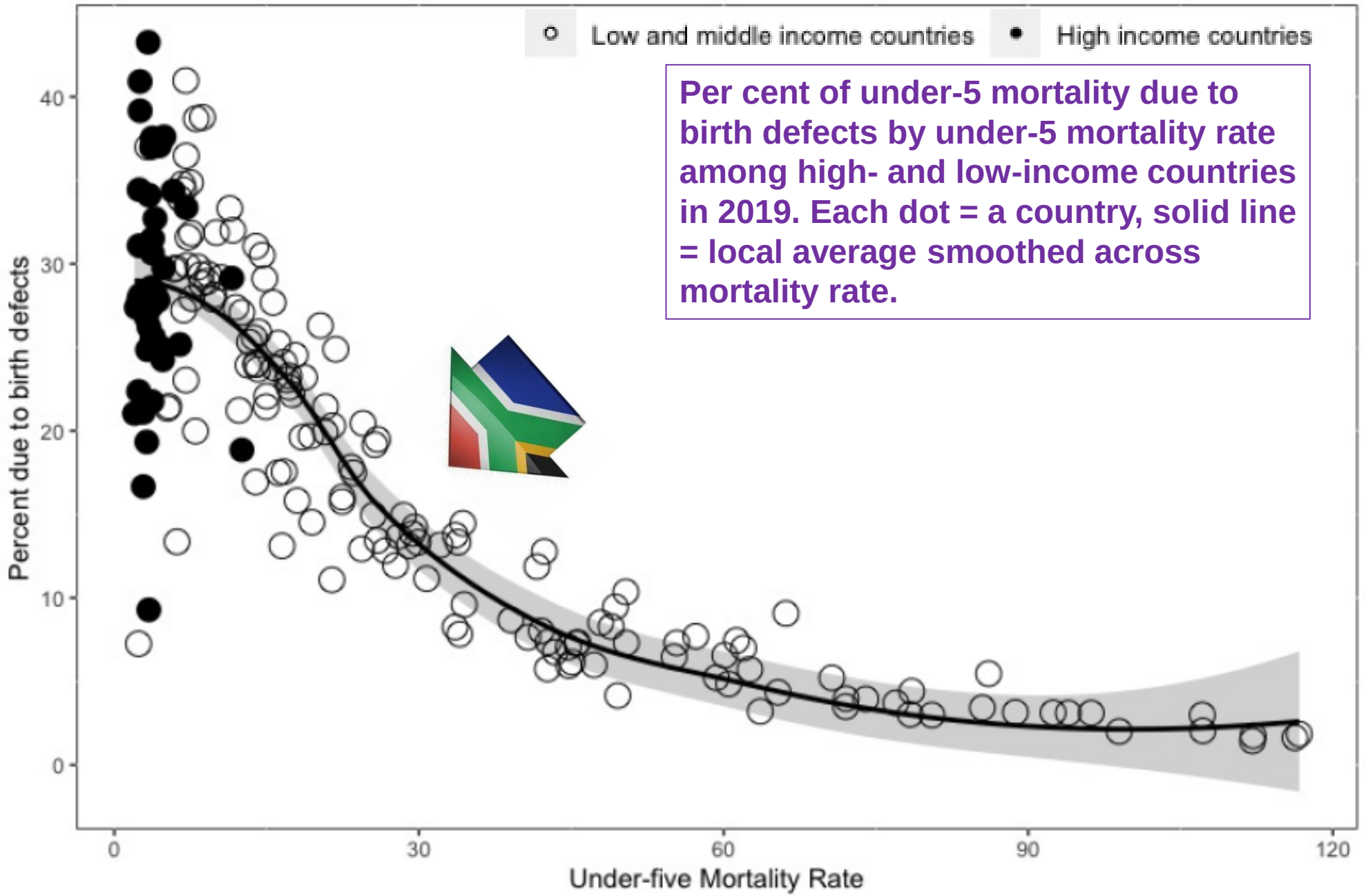
Global context and trends (Perin et al 2023)

- CDs are now among top 5 causes of under-5 deaths in MIC;
- Overall mortality rate declined modestly: **3.4 → 2.9 per 1,000 live births**
- But **share of all under-5 deaths increased**: **4.6% → 7.6% globally**

Less children are dying overall but proportionally more are dying from CDs

- In neonates (0–27 days), CDs now account for **~10% of deaths (survival issue);**
- **404,000 CD related under-5 deaths in 2019** (↓ from 452,600 in 2000);
- **LMIC mortality rate >2× higher** than in high-income countries (3.0 vs 1.3 per 1,000);
- In HICs, CD account for a **much larger share of child deaths** because infectious causes are low; (i.e. U5MR >25 per 1000 5% due to CD; U5MR <25 per 1000 25%);

As overall child mortality declines, % attributable to CDs rises sharply.



Per cent of under-5 mortality due to birth defects by under-5 mortality rate among high- and low-income countries in 2019. Each dot = a country, solid line = local average smoothed across mortality rate.

Source: Perin et al 2023

South African Context

- 1.2 million km^2 over 9 provinces
- **Population:** 63m; 35% ≤20 years; 10% ≥ 60 (2021)
- **Urbanised:** 68% (2021)
- **Life Expectancy:** 57.3-67.6 (♂) 64.2-72.2 (♀)
- **Annual Births:** approx. 1 million P/A
- **Fertility Rate:** 2.41 (2024)
- **Mothers Age:** 14.4% ≤ 20 yrs; 18.5% ≥35 yrs (2025)
- **U5MR:** 28/1 000 LB (2021)
- **IMR:** 21/1 000 LB (2021)
- **NNMR**:12/1 000 LB (2021)
- **HIV**: 13.7% = approx. 8m (2022)
- **Dual healthcare**: 86% public:16% private

38% global U5 deaths due to CA occur in SSA (2019)

Adapted from: https://commons.wikimedia.org/wiki/File%3AMap_of_South_Africa_with_English_labels.svg

Stats SA 2025; Dorrington et al (RMS) 2021

South African Context: what does the data say?

- Expected prevalence: 3–6% of live births;
- An estimated **30,000–60,000 affected infants born annually in SA;**
- CDs among top causes of neonatal mortality in SA (crosscutting);
- As infectious mortality declines, proportional contribution of CD increases;
- Current surveillance in SA limited to major CD categories only & underestimating true burden by >90% (Lebese et al 2016);
- Under-reporting at facility level & limited linkage between birth record (no EHR), perinatal mortality data, genetic services; poor post-discharge follow-up; inconsistent ICD coding (Patrick et al 2018);

Implication: Underestimation of the burden of CD → leads to under-prioritisation in planning and budgeting – so those affected/at risk **do not received required care.**

Epidemiological Transition: Why CDs Matter Now

- ↓ HIV-related vertical transmission - HIV deaths no longer dominate neonatal mortality (public health success);
- ↓ Overall infectious causes of death - space opens for other causes;
- ↑ Relative proportion of deaths due to congenital causes - persistent, non-declining burden;
- ↑ Maternal age shifts - changes risk profile;
- Persistent high FASD burden – large preventable contribution;
- Resurgence of congenital syphilis (stillbirth/neonatal mortality) – preventable condition;

Epidemiological transition is shifting neonatal mortality from infectious to congenital drivers.

Economic and Health System Impact

- **High cost, complex conditions**: often require scarce specialist services: Tertiary surgery (cardiac, NTD, OFC etc), NICU and repeat admissions, long-term care and rehabilitation, special education, social & disability support.
- **System wide fiscal pressure** – costs extend beyond health including education, employment, social development.
- **Household economic impact:** Entrenched intergenerational poverty due to catastrophic out-of-pocket costs (transport, caregiving time), reduced parental employment (especially mothers), long-term income loss.
- **Productivity and human capital effects** – without required early care leads to preventable disability, reduced education, lower lifetime earnings, reduced labour market participation versus mainstream schooling, increased employability/contribution, reduced dependency;
- **Prevention is highly cost effective** – costs a fraction of lifelong treatment e,g, FAFF, syphilis screening, preconception/ANC – but have to care for those affected;
- **Cost of inaction** – unaddressed (undiagnosed, uncounted, untreated) CDs lead to avoidable mortality and disability lost productivity, increased long-term state expenditure.

***"Care is the absolute, prevention the ideal"* (Christianson et al 2000).**

Current Service Landscape

- **Highly centralised service model** - Genetic services concentrated in tertiary centres, urban areas, referral dependent model;
- **Severe workforce constraints -** limited training pipeline & heavy caseloads;
- **Significant geographic inequity** - uneven provincial access, rural/urban;
- **Minimal public NBS** – missed opportunities for early detection/intervention;
- **Weak integration into PHC** – limited genetic literacy inconsistent ANC risk identification, weak referral pathways;

CDs not yet fully mainstreamed into maternal and child health platform.

Population & numbers of practicing medical geneticists (MG) & genetic counsellors (GC) in South Africa, 2024

Overall:

- 8% recc. public MG
- 2% recc. public GC
- 20% recc. Private MG
- Publicly practicing MG & GC in WC and GP ONLY
- 22% recc. Private GC
- 6/9 provinces have no MG
- 5/9 provinces have no MG

Biggest challenge is emigration!

Source: Gomes et al 2024

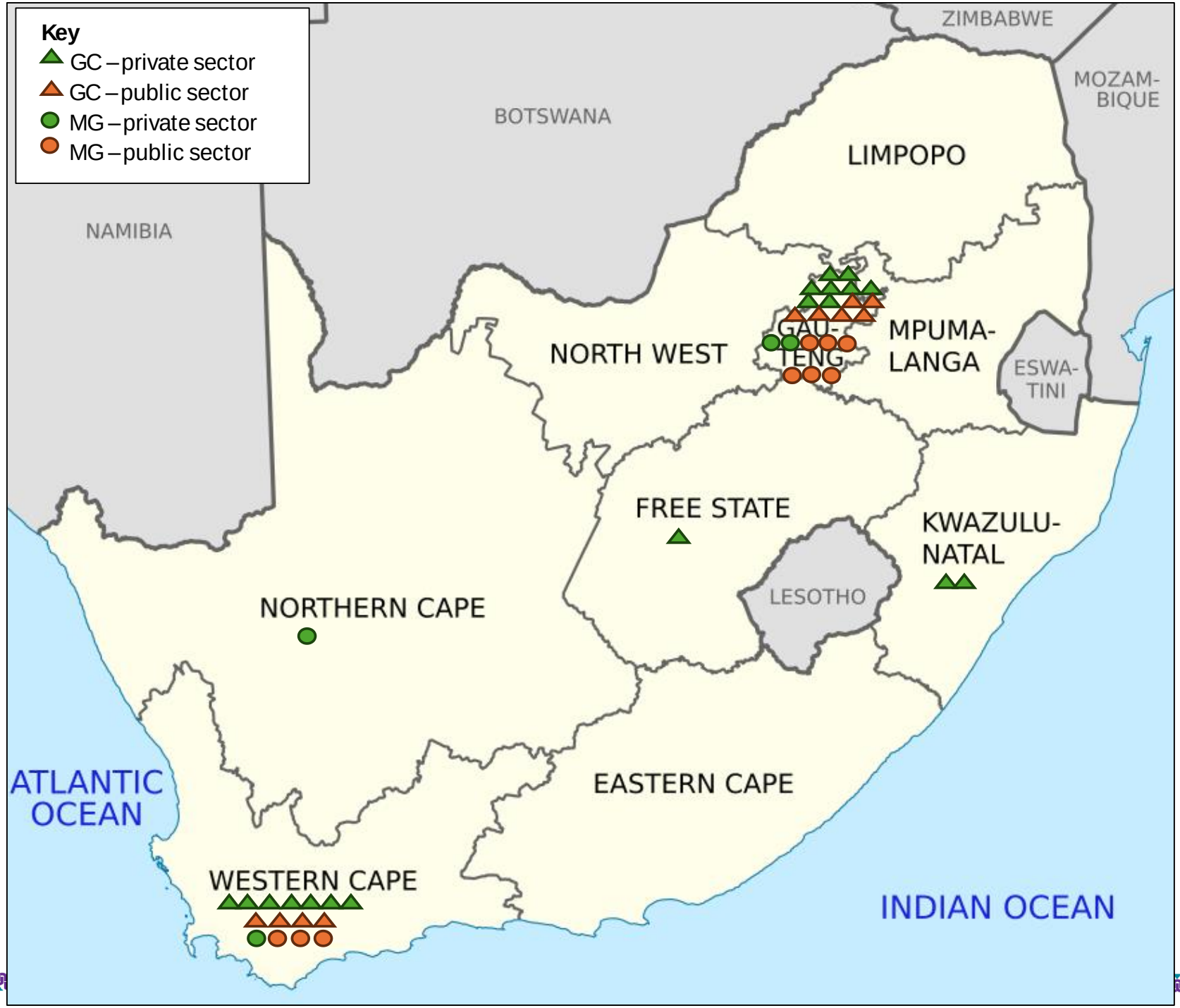


image: https://www.statista.com/statistics/1112169/total-population-of-south-africa-by-province/)

Implications - What should we focus on?

1. Strengthen Surveillance:

- National CD registry
- Standardised reporting
- Integration with DHIS

2. Prevention at PHC Level:

- Folic acid supplementation policy strengthening
- Alcohol reduction strategies (FASD)
- Syphilis screening enforcement
- Diabetes control before/during pregnancy

3. Preconception and Early Antenatal Care:

- Promote early ANC booking (<12 weeks)
- Preconception counselling & risk screening tools

4. Expand Genetic & Diagnostic Capacity:

- Training of PHC staff
- Tele-genetics
- Provincial referral pathways

5. Newborn Screening & Early Intervention:

- Scale pilot NBS programmes
- Early surgery referral pathways
- Linkage to rehabilitation services

Global Momentum

Global Initiatives:

- **Sustainable Development Goal 3 Ensure Healthy Lives and Promote Wellbeing for All and at All Ages – 2030 targets.**
- **International Rare Diseases Research Consortium (IRDiRC)** – global collaboration on RD research & diagnostics.
- **Rare Diseases International (RDI)** – patient-led global advocacy, specifically:
  - **RDI memorandum with WHO -** shape international RD policy on rare diseases and strengthen health systems, advance UHC.
  - **Collaborative Global Network for Rare Diseases (RDI)** –platform for registries, data, and international cooperation.
  - **RDI Essential Medicines List (EML) Initiative** – advancing inclusion of RD medicines in national & global EMLs.

World Health Assembly (WHA) Resolutions:

- **WHA63.17 (2010)** – Birth defects: reducing the impact of preventable birth defects.
- **WHA74.2 (2019)** – Political declaration of the high-level meeting on universal health coverage.
- **WHA76.13 (2021)** – Addressing the challenges of persons living with a rare disease and their families.
- **WHA76.19 (2023)** – Accelerating efforts for preventing micronutrient deficiencies and their consequences.
  WHA77.5 (2024) – Accelerate progress towards reducing maternal, newborn and child mortality to achieve SDG targets 3.1 & 3.2.
- **WHA78.11 (2025) – Rare diseases: a global health priority for equity and inclusion.**

World Health Organization

WHO member states are required to report progress on WHA's

NWU® | NMP | technology innovation AGENCY

Concluding Remarks

- CDs are predictable and measurable;
- Many are preventable;
- They are a persistent contributor to neonatal/child mortality and disability;
- As infectious deaths decline CDs account for a growing share of remaining neonatal deaths;
- Without integrating CD into routine maternal and child health services, further reductions in neonatal mortality will stall.

If South Africa is serious about reducing neonatal mortality below SDG targets, CDs must move from the margins to the mainstream of maternal and child health policy.

Thank you – questions?

Helen.Malherbe@nwu.ac.za
https://natural-sciences.nwu.ac.za/human-metabolomics
https://nationalmetabolomicsplatform.co.za

Key References

1. Christianson AL, Venter PA, Modiba JH, Nelson MM. Development of a primary health care clinical genetic service in rural South Africa – The Northern Province experience,1990–1996. Community Genetics. 2000; 3(2):77–84.
2. Czeizel AE, Intody Z, Modell B. What proportion of congenital abnormalities can be prevented? BMJ. 1993;306(6876):499-503.
3. Dorrington R, Bradshaw D, Laubscher R, Nannan N. Rapid Mortality Surveillance Report 2019 & 2020. Cape Town: South African Medical Research Council, 2021.
4. Gomes MCM, Gomes B, Bailly C, McKerrow N, Christianson AL, Malherbe H. A capacity audit of medical geneticists and genetic counsellors in South Africa, 2024 – A national crisis. Genes. 2024;15((9)):1173.
5. Lebese V, Aldous C, Malherbe HL. South African congenital disorders data, 2006 - 2014. S Afr Med J. 2016;106(10):992-5.
6. Malherbe H, Aldous C, Woods D, et al. The contribution of congenital disorders to child mortality in South Africa. In: Padarath A, King J, Mackie E, et al., eds. South African Health Review 2017. 19th ed. Durban: Health Systems Trust 2016:137-52.
7. Malherbe HL, Bonham JR, Carrihill M, Chetty KS, Conradie EH, Dercksen M, et al. Newborn screening in South Africa: the past, present and plans for the future. Rare Dis Orphan Drugs J. 2024;3(7):11.
8. Malherbe HL, Modell B, Blencowe H, Strong KL, Aldous C. A review of key terminology and definitions used for birth defects globally. J Community Genet. 2023;14:241-62.
9. Malherbe H & Odendaal (2025). Breaking Down the Burden of Rare Diseases in South Africa. Innovative Pharmaceutical Association South Africa, Johannesburg: South Africa.
10. National Department of Health. Clinical Guidelines for Genetic Services. Pretoria: National Department of Health, 2021.
11. Patrick M, Malherbe H, Stephen C, Woods D, Aldous C. Congenital disorders in South Africa: A review of Child Healthcare Problem Identification Programme (Child PIP) mortality data, 2005-2017. S Afr Med J. 2018;108(8):6.
12. Perin J, Mai CT, De Costa A, Strong K, Diaz T, Blencowe H, et al. Systematic estimates of the global, regional and national under-5 mortality burden attributable to birth defects in 2000–2019: a summary of findings from the 2020 WHO estimates. BMJ open. 2023;13(1).
13. Statistics South Africa. Statistical Release P0305. Recorded live births 2024. Pretoria: Stats SA, 2025.
14. Statistics South Africa. Statistical Release P0302. Mid-year Population Estimates 2025. Pretoria: StatsSA, 2025.