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Congenital disorders and medical genetic services in South Africa

Helen Malherbe
PhD Candidate
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Outline

- Definitions
- Overview of SA situation
- Epidemiological transition
- Legislation & policy
- Medical genetic services
- Conclusions

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Definitions

Structural and chromosomal fetal anomalies

Inherited disorder

Congenital abnormalities

Genetic disorders

Birth defect

Chromosomal defect

Fetal abnormalities

Chromosomal abnormalities

Familial diseases

Congenital anomalies

Congenital malformations

Fetal anomaly

Congenital infection

Congenital disorder

Genetic disease

Chromosomal abnormality

Definitions

Congenital disorders = birth defects

"Abnormalities in structure or function, including metabolism, which are present from birth" (WHO, 2006)

- *Obvious at birth, in childhood, or later in life*
- *Genetic/partially genetic, environmental, combination of these or unknown factors*
- *Congenital disorders include rare diseases*
- *Congenital anomalies ≠ congenital disorders (CDs)*
- *Congenital malformations are also a sub-set of CDs*

Definitions

- Global confusion in terminology
- Prevents data sharing & comparison
- Impacts rankings e.g. GBD 2013
- ICD-10 system: Chapter XVII excludes 40%
- WHO 2006 agreement
- Inequivalent terms still used
- Sub-sets reported as totality
- Underreporting & inaccurate estimate

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ICBD Pledge

"Building consensus on and widespread use of a ***standardized definition of congenital disorders****.... to facilitate data comparison and ensure that the contribution of congenital disorders to the burden of disease is comprehensively represented."*

Less is more?

- CDs a global issue (7%)
- Unequally distributed
- >90% occur in developing countries
- Leading cause of death in high income countries
- Not yet recognized in developing countries
- Undiagnosed, misdiagnosed & underreported

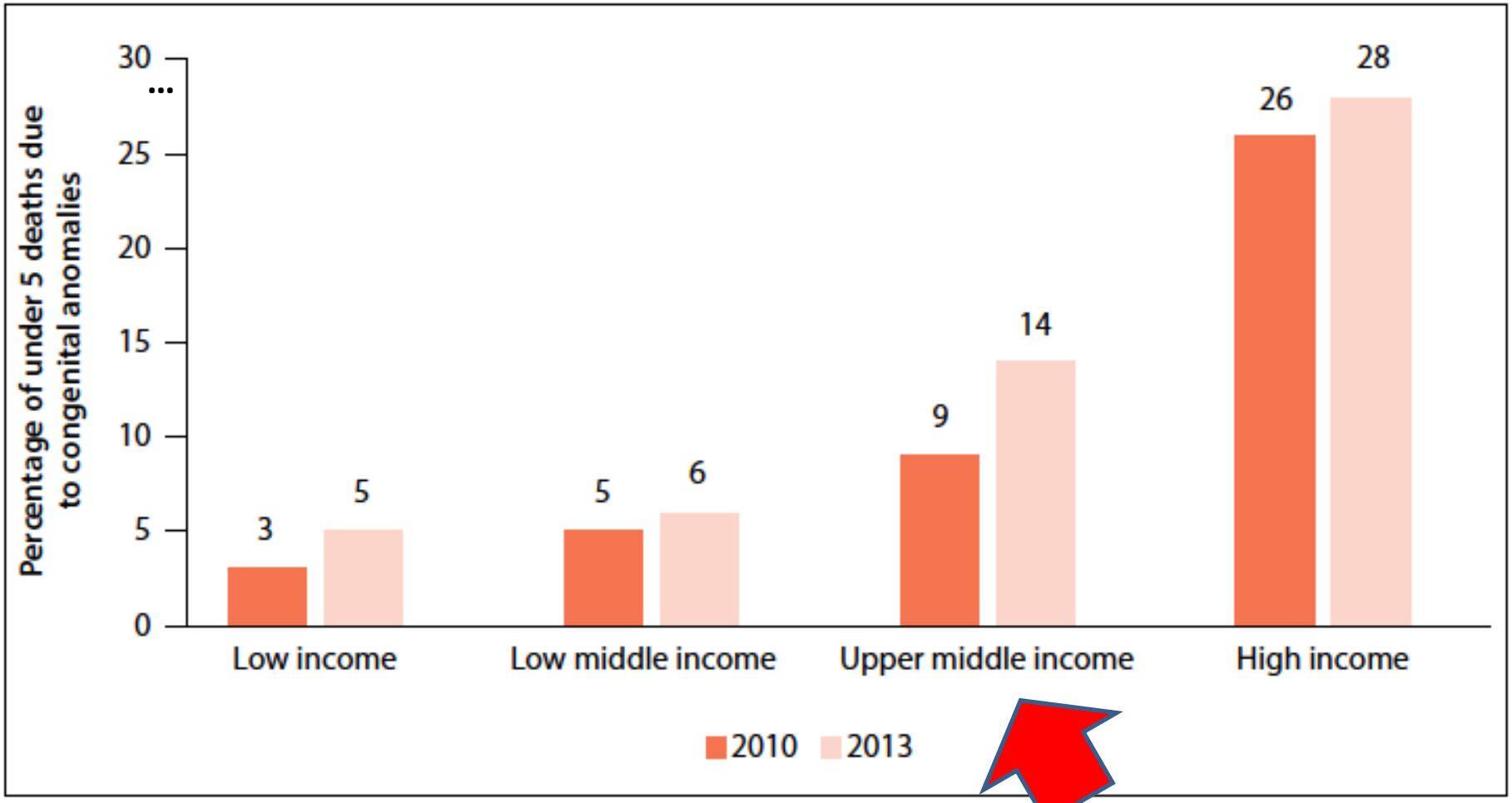

Fig. 1. Comparison of the percentage of under-five deaths resulting from congenital anomalies for World Bank Country Classifications. [9]

Source: Malherbe et al, SAJBL, May 2016 9(1)

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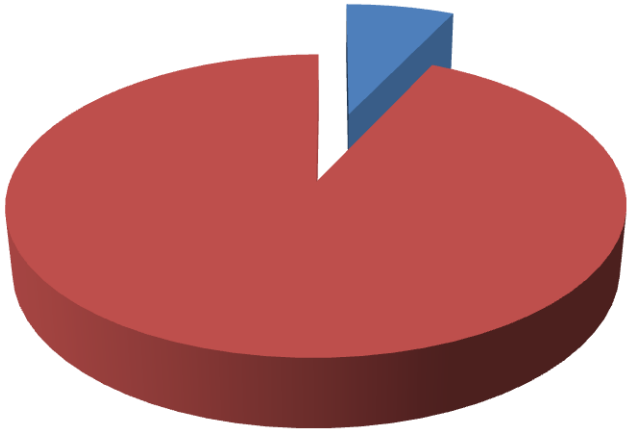
South Africa

Estimated annual congenital disorders in South Africa



1 in every 15 live births (6.8%) affected by a congenital disorder

Of which:

- **80.5% genetic/partially genetic**
- **19.5% teratogens (FAS major factor)**

National surveillance underreporting by 98% (Lebese et al 2016)

Malherbe et al, 2015. S Afr Med J 2015;105(3):186-188.

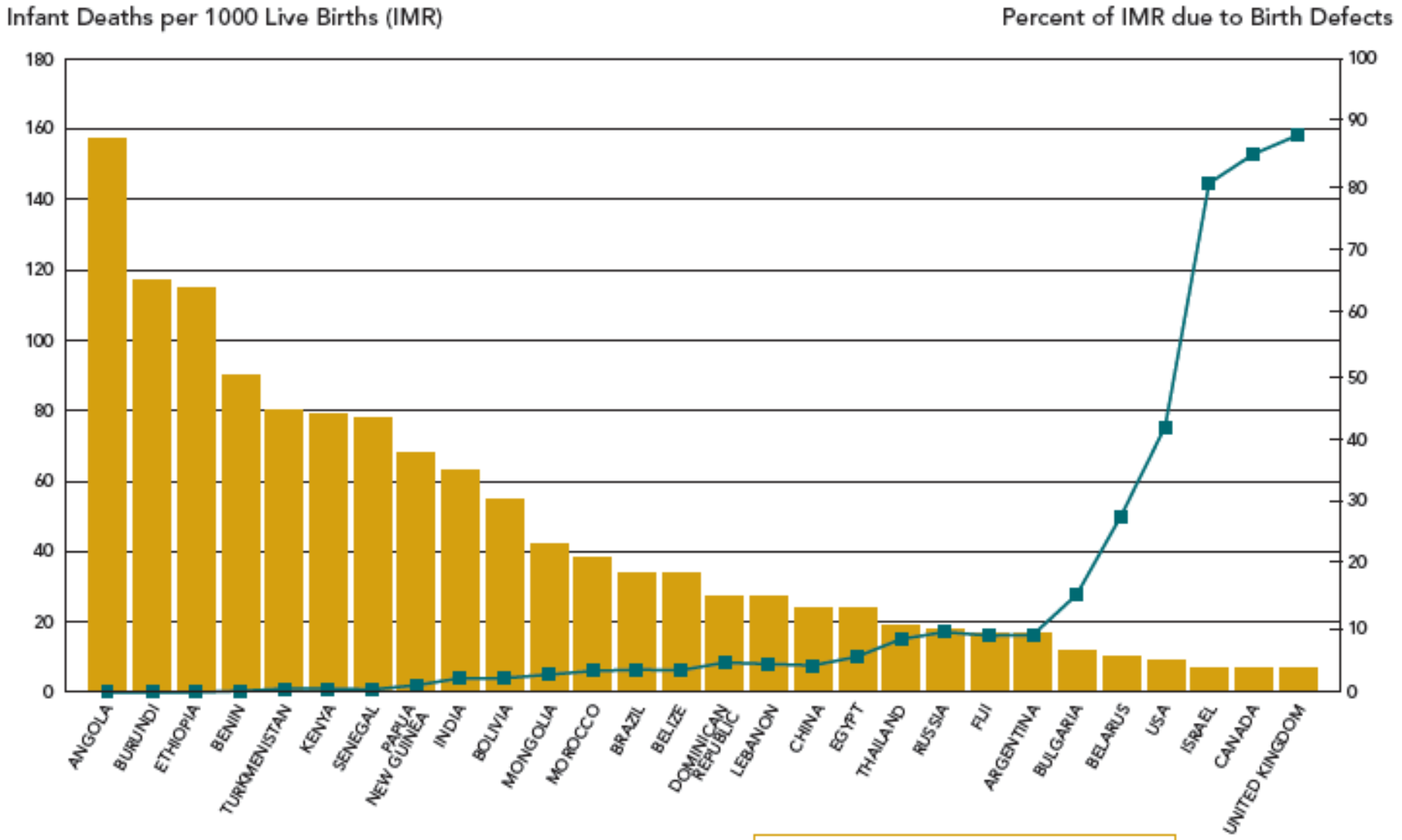
Epidemiological Transition

"The change in population health statistics and pattern of diseases of a country/region, consequent on change in socio-economic, education, infrastructure or heath care development."

Omran's Model

- Infant & child mortality decrease
- Life expectancy at birth (longevity) rises
- Communicable diseases controlled/eradicated
- Non-communicable & degenerative diseases emerge
- High income countries: **classic transition**
- MLIC: **protracted transition** (double burden)
- Proportion of CD related deaths increases (**RELATIVE** versus **ABSOLUTE**)

FIGURE 1. RELATIONSHIP BETWEEN INFANT MORTALITY RATE (IMR) AND PERCENTAGE OF INFANT DEATHS DUE TO BIRTH DEFECTS IN THE ABSENCE OF KNOWN PREVENTIVE SERVICES BY COUNTRY, 2004



Sources: UN 2006; Bernadette Modell, personal communication, 2008.

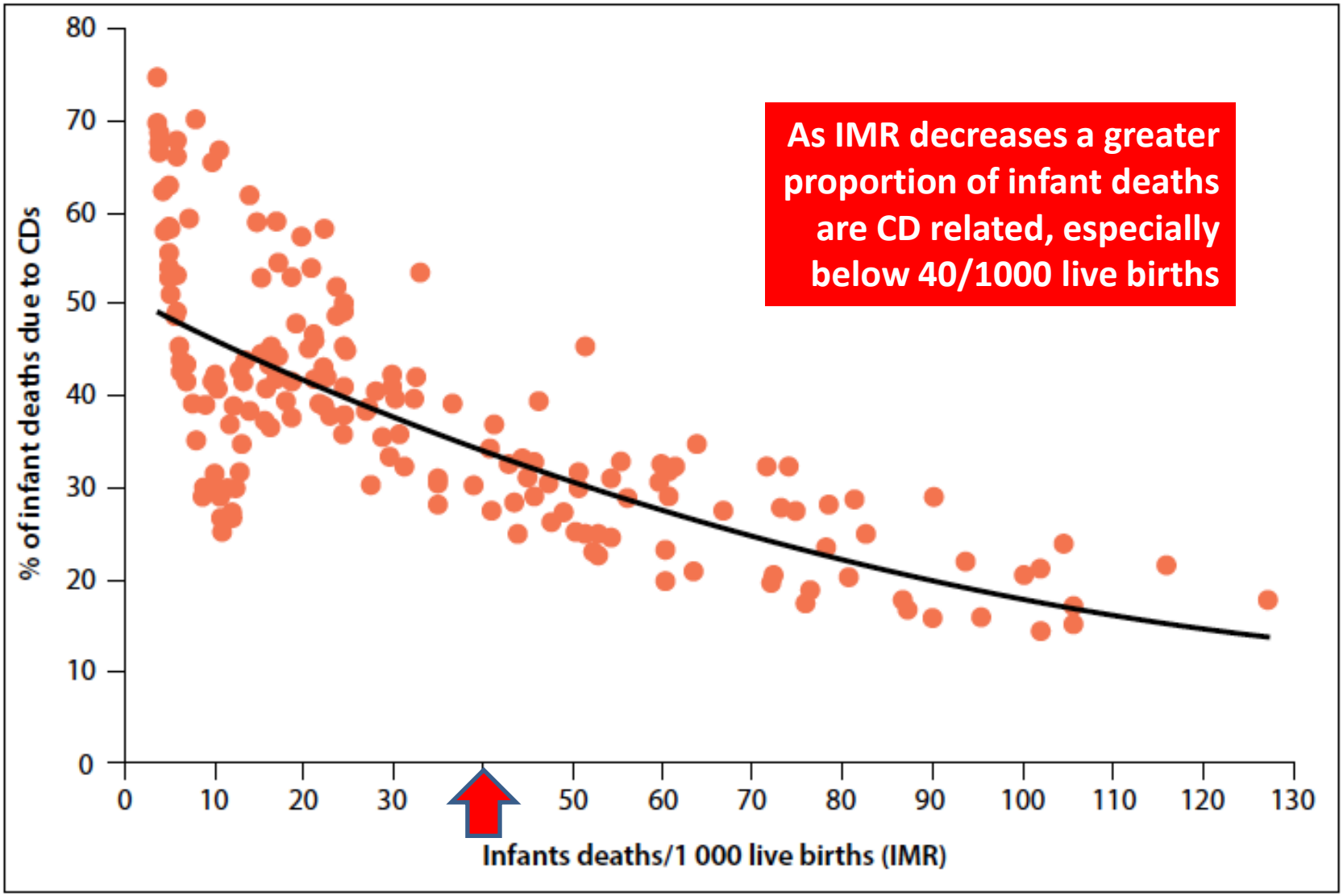
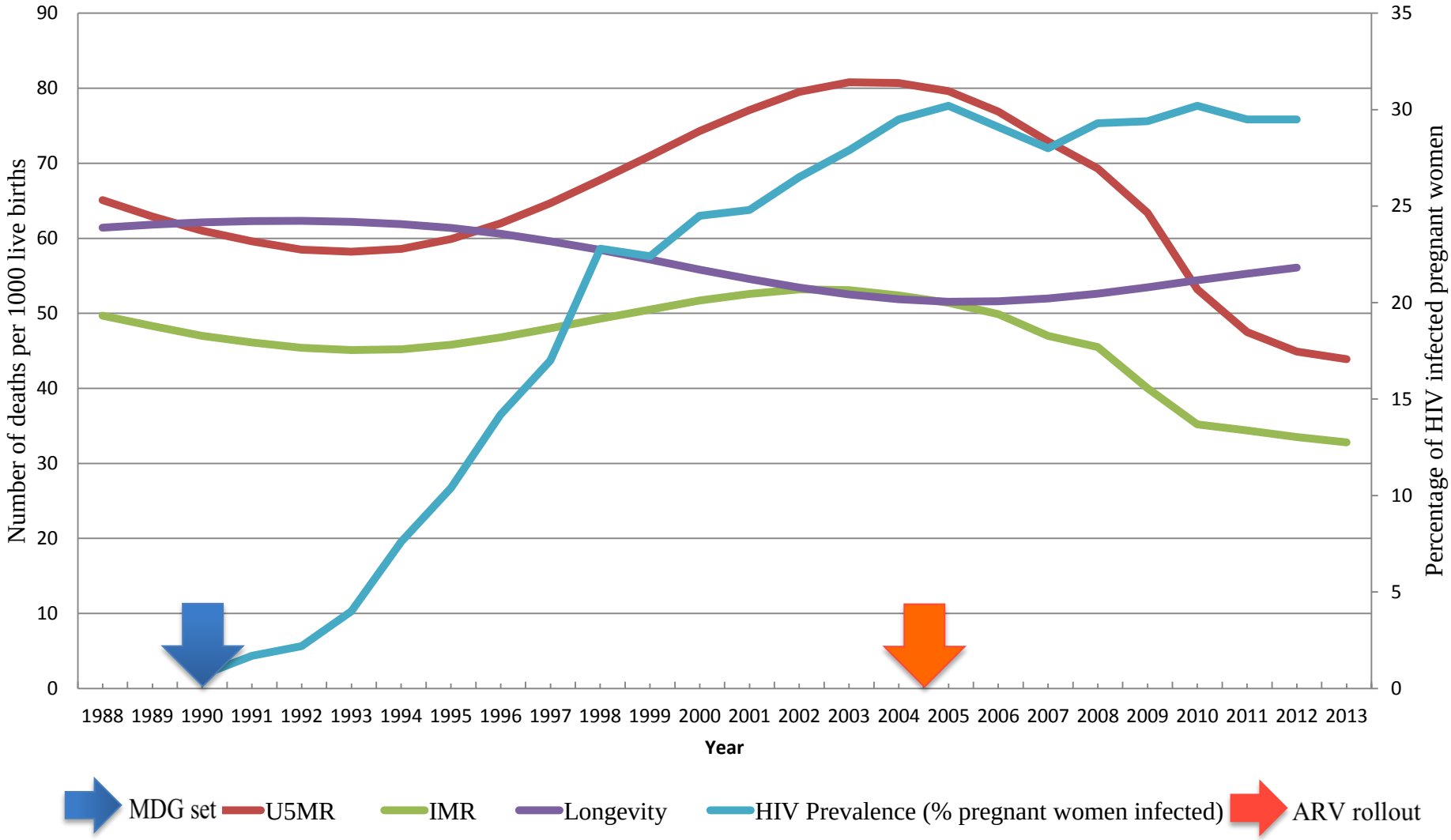


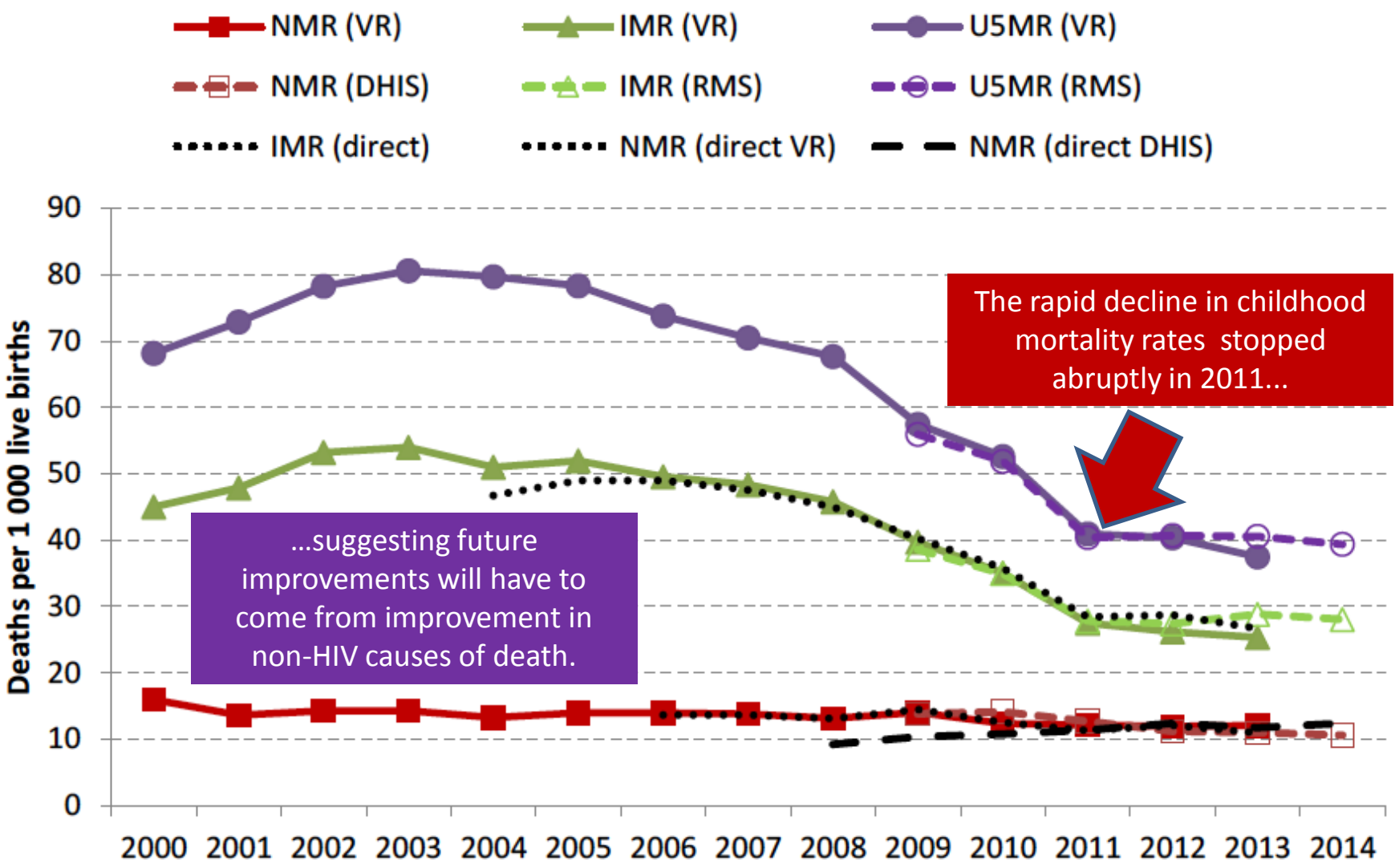
Fig. 2. Relationship between infant mortality and percentage of infants dying from CDs, based on global country figures.[9]

Source: Malherbe et al, SAJBL, May 2016 9(1)

Figure 1. Epidemiological transition in South Africa over the past 25 years



Malherbe et al, 2015. S Afr Med J 2015;105(3):186-188.



Under-5 Mortality rate (U5MR) and Infant Mortality Rate (IMR) From VR/RMS And Neonatal Mortality Rate (NMR) From VR/DHIS, 2000-¡©-2014 (Adjusted)

Source: MRC Rapid Mortality Surveillance Report 2014

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Global Political Will

"attainment of MDG4 (Reduce child mortality) will require accelerated progress in reducing neonatal mortality including prevention and management of birth defects"
(63rd World Health Assembly, Resolution 63.17, 2010)

World Health Organization

SA Constitution: Bill of Rights

- Equality, human dignity & fundamental right to life
- Right to access to healthcare services, including reproductive (Section 27)
- Every child has the right to basic health care services (Section 28)
- Progressive realisation…

National Health Act

Section 21:

The Director General of the National Department of Health **must**, in accordance with national health policy, B) issue and promote adherence to, norms and standards on health matters including vii) **genetic services**

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National Legislation

- Constitution of the Republic of South Africa
- Health Professions Act
- National Health Act
- Choice on Termination of Pregnancy Act
- NHLS Act
- Mental Health Care Act
- The Nursing Act
- The Children's Act
- The Social Assistant Act

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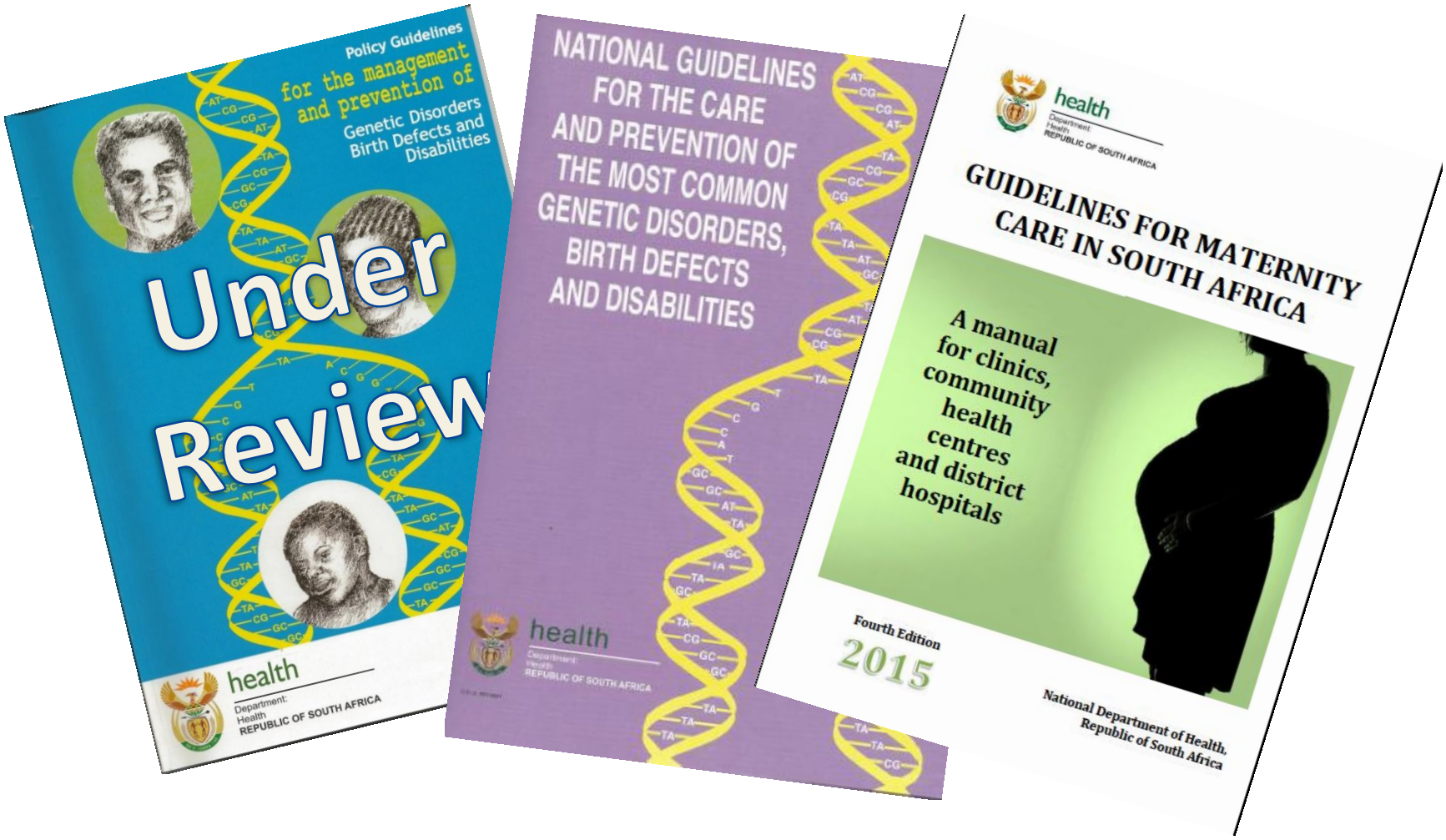
Policy



Medical genetic services

- *"To help people with a genetic disadvantage to live and reproduce as normally and responsibly as possible"*
- "Best possible patient care in prevailing circumstances and prevention by appropriate interventions"
- "Care is an absolute. Prevention the ideal" (Christianson, 2000)
- Primary, secondary & tertiary prevention (care)

Myths

- Birth defects are not an issue…
- “Nothing can be done, they’re going to die anyway”
- Its too expensive!
- There’s no capacity or infrastructure for the high-tech, vertical programmes needed
- Funding for CDs will take funds away from other priority healthcare issues.

Truth

- CDs are underreported by 98% in SA
- 70% of CDs can be prevented, cured or ameliorated (Czeizel, 1993)
- Not all interventions are high tech/expensive
- There is a cost to ‘no care’
- Incorporate/integrate into existing PHC programmes

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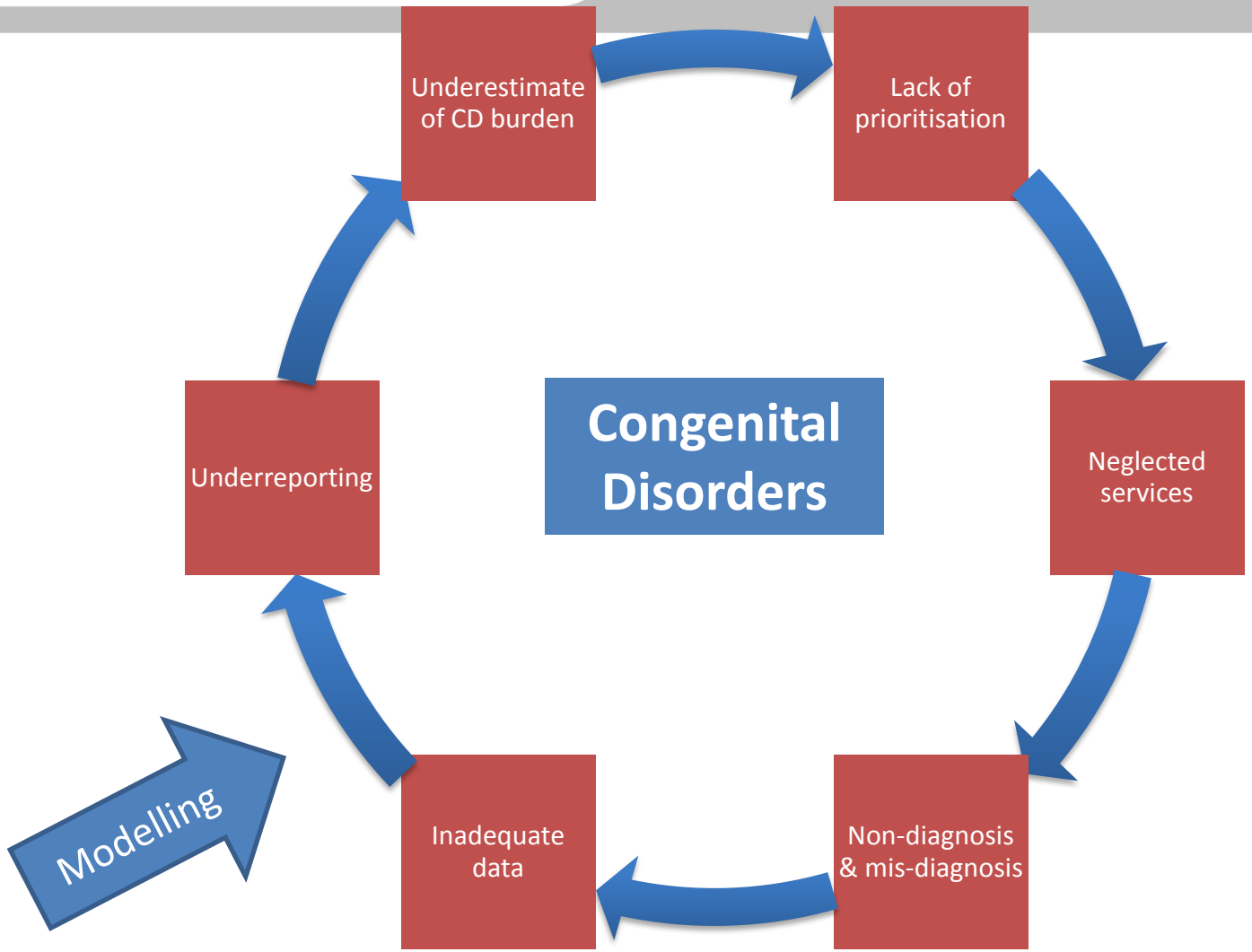
Historically

- Academic depts & medical schools since 1970s
- Few community based services
- Access limited to mainly urban areas with some outreach into rural areas
- Genetic nurses NW integrated into primary care
- Implementation of 2001 policy limited
- Medical Genetics Education Programme (MGEP)
- Competing health priorities

Observations

- Legal framework but **poor implementation**
- Focus on reducing child mortality excluding CDs
- CDs not contextualised as an NCD
- Contribution of CDs not acknowledged or tackled **comprehensively**
- People with disability (esp children) are the most marginalised group
- Data is starting to emerge (**WC neonates**)

The cycle


Underestimate of CD burden
Lack of prioritisation
Neglected services
Non-diagnosis & mis-diagnosis
Inadequate data
Underreporting
Congenital Disorders
Modelling

A comparison of medical genetics services capacity in 2001 and 2015

	Recommended*	2001		2015	
Category	No./ratio (x= 46 127m)†	No.	Ratio (x= 44 820m)†	No.	Ratio (x=54 937 m)§
Medical geneticists	20/1 per 2 m	4	1 per 11.2 m	11¶	1 per 4.9 m
Genetic counsellors	80/1 per 580 000	20	1 per 2.2 m	7.5∏	1 per 7.3 m
Medical scientists/ technologists	100/1 per 450 000	50	1 per 900 000	26**	1 per 2.1 m

*Department of Health. Strategic Framework for the Modernisation of Tertiary Hospital Services.
†Statistics South Africa. South African Statistics 2014.
‡Department of Health. Policy Guidelines for the management and prevention of genetic disorders, birth defects and disabilities. 2001
§Statistics South Africa. Mid-Year Population Estimates 2015.
¶ Prof Amanda Krause, personal communication.
∏Plus seven genetic counsellors in private practice (Dr Tina Wessel & Shelley McCaulay personal communications).
**NHLS medical scientists only (Prof Himla Soodyall, personal communication).

Source: Adapted from Malherbe *et al*, South African Health Review 2016

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Conclusions

- "*We have good laws: the constitution, legislation, guidelines and policy – but the services are being lost in translation*" (M. Christianson, 2007)
- Genetic services have declined in the past decade
- Failure to recognise disease burden of CDs
- Well beyond the point (IMR 40/1000) when services should be implemented to reduce child mortality
- CDs will emerge as a leading cause of child death and disability
- Buried under "quadruple" burden of disease in SA
- Imperative to respond to WHA 63.17 and prioritize CDs in tandem with low-hanging fruits

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Forum: building unity in the human genetics community
Simonsburg: 17:15

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Thank you

Madeleine Eva Malherbe
14 Oct - 24 Dec 2004

"no single life is better than another"