Congenital disorders and medical genetic services in South Africa

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Outline

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

Structural and chromosomal fetal anomalies
  Congenital abnormalities
  Birth defect
  Chromosomal abnormalities
  Familial diseases

Inherited disorder
  Genetic disorders
  Chromosomal defect
  Fetal abnormalities

Congenital anomalies
  Congenital malformations
  Genetic disorder
  Fetal anomaly

Congenital infection
  Chromosomal abnormality

Genetic disease

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INSPIRING GREATNESS
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
“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.\textsuperscript{[9]}

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

Estimated annual congenital disorders in South Africa

Of which:
• 80.5% genetic/partially genetic
• 19.5% teratogens (FAS major factor)

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

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

“The change in population health statistics and pattern of diseases of a country/region, consequent on change in socio-economic, education, infrastructure or health 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

Infant Deaths per 1000 Live Births (IMR)

Percent of IMR due to Birth Defects

Sources: UN 2006; Bernadette Modell, personal communication, 2008.
As IMR decreases a greater proportion of infant deaths are CD related, especially below 40/1000 live births.

Source: Malherbe et al, SAJBL, May 2016 9(1)
Figure 1. Epidemiological transition in South Africa over the past 25 years

The rapid decline in childhood mortality rates stopped abruptly in 2011...

...suggesting future improvements will have to come from improvement in non-HIV causes of death.

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
“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)
• 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...
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**
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
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)
• Birth defects are not an issue...
• “Nothing can be done, they’re going to die anyway”
• It’s 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
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
• 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

INSPIRING GREATNESS
A comparison of medical genetics services capacity in 2001 and 2015

<table>
<thead>
<tr>
<th>Category</th>
<th>Recommended*</th>
<th>2001</th>
<th>2015</th>
<th></th>
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<tr>
<td></td>
<td>No./ratio</td>
<td>No.</td>
<td>Ratio</td>
<td>No.</td>
</tr>
<tr>
<td></td>
<td>(x=46 127m)†</td>
<td></td>
<td>(x=44 820m)†</td>
<td></td>
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<tr>
<td>Medical geneticists</td>
<td>20/1 per 2 m</td>
<td>4</td>
<td>1 per 11.2 m</td>
<td>11†</td>
</tr>
<tr>
<td>Genetic counsellors</td>
<td>80/1 per 580 000</td>
<td>20</td>
<td>1 per 2.2 m</td>
<td>7.5Ⅱ</td>
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<tr>
<td>Medical scientists/technologists</td>
<td>100/1 per 450 000</td>
<td>50</td>
<td>1 per 900 000</td>
<td>26**</td>
</tr>
</tbody>
</table>

‡Department of Health. Policy Guidelines for the management and prevention of genetic disorders, birth defects and disabilities. 2001
¶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
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
Forum: building unity in the human genetics community
Simonsburg: 17:15
“no single life is better than another”

Madeleine Eva Malherbe
14 Oct - 24 Dec 2004