NHI in the context of Rare Diseases

Rare Diseases South Africa

Agenda

- Introduction to Rare Disease South Africa NPC.
- Introduction to Mrs Kelly du Plessis and Dr Helen Malherbe.
- Context of Rare Diseases in South Africa.
- Discussion points with relation to NHI Bill.
- Universal Health Care.
- Closing and Thank You.

Discussion Areas

Relative to 2018 feedback:

- Diagnostic Services.
- PMB criteria.
- Issue of progressive realization.
- Lack of prioritization of rare diseases as a health care issue.
- Lack of human resources in medical genetics.
- Referral services.

Discussion Areas

Relative to 2019 feedback:

- Health system revolution through legislation.
- The inclusion of civil society and health service users in all decisions and processes.
- Governance under NHI.
- Emergency Medical Services.
- Transparency.
- Financing NHI.
- Competition law and the NHI;
- Implementation of the recommendations of the Health Market Inquiry in the interests of NHI.

Who is Rare Diseases SA?

- Rare Diseases South Africa (RDSA) is a registered non-profit (NPO) and public benefit organization (PBO).
- RDSA represents patients affected by rare diseases and congenital disorders, support groups aimed to assist this patient group, as well as healthcare practitioners, researchers and academics working towards improving the quality of life of these patients.
- RDSA currently has a membership base of over 5000 members.

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aims and objectives of RDSA

our aims

- Increase awareness around rare conditions.
- Provide support and practical aid to patients and families impacted by rare conditions and congenital disorders.
- Establish a network between all partners with the ability to prevent, affirm, treat and improve the quality of life of affected individuals.

our vision

A South Africa where those impacted by rare diseases and congenital disorders access life-saving treatment and supportive care for improved quality of life.

our mission

Bridging the gap to improved quality of life for those impacted by rare diseases and congenital disorders through advocacy and empowerment.

our principles

Equality, Care, Dignity and Empowerment.

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Rare Diseases – Global Context

Rare Diseases South Africa

Only **5%** of rare diseases have an FDA approved drug

Approximately **30%** of children with a rare diseases will not live to see their **5th** birthday

More than **350 million** people are affected globally

Over **7000** Rare Diseases identified

80% of Rare Diseases are genetic

50% of people affected by Rare Diseases are children

South African Context

Rare Diseases South Africa

Globally Rare Diseases impact 350 million people

Quality of life for people with Rare diseases is not dependent on severity of the illness, but rather on:

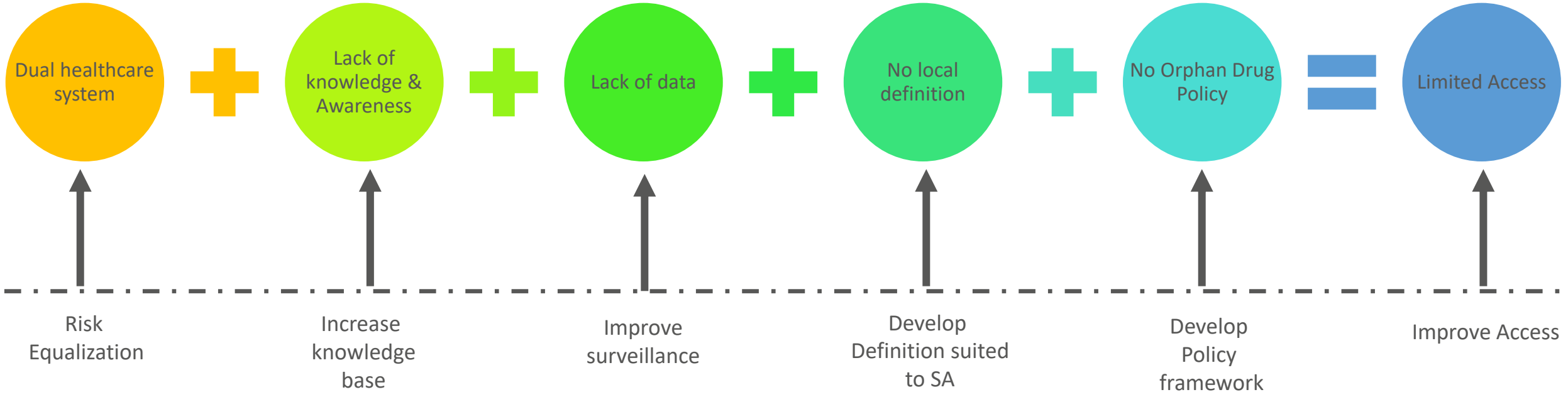
- Availability of treatment
- Health Systems
- Broader Social Support
- Access to Supportive care

(Rajmil et al. 2010)

Estimated 3.7 million South Africans are impacted by Rare Diseases based on modelled data.

Rare Diseases South Africa

Challenges



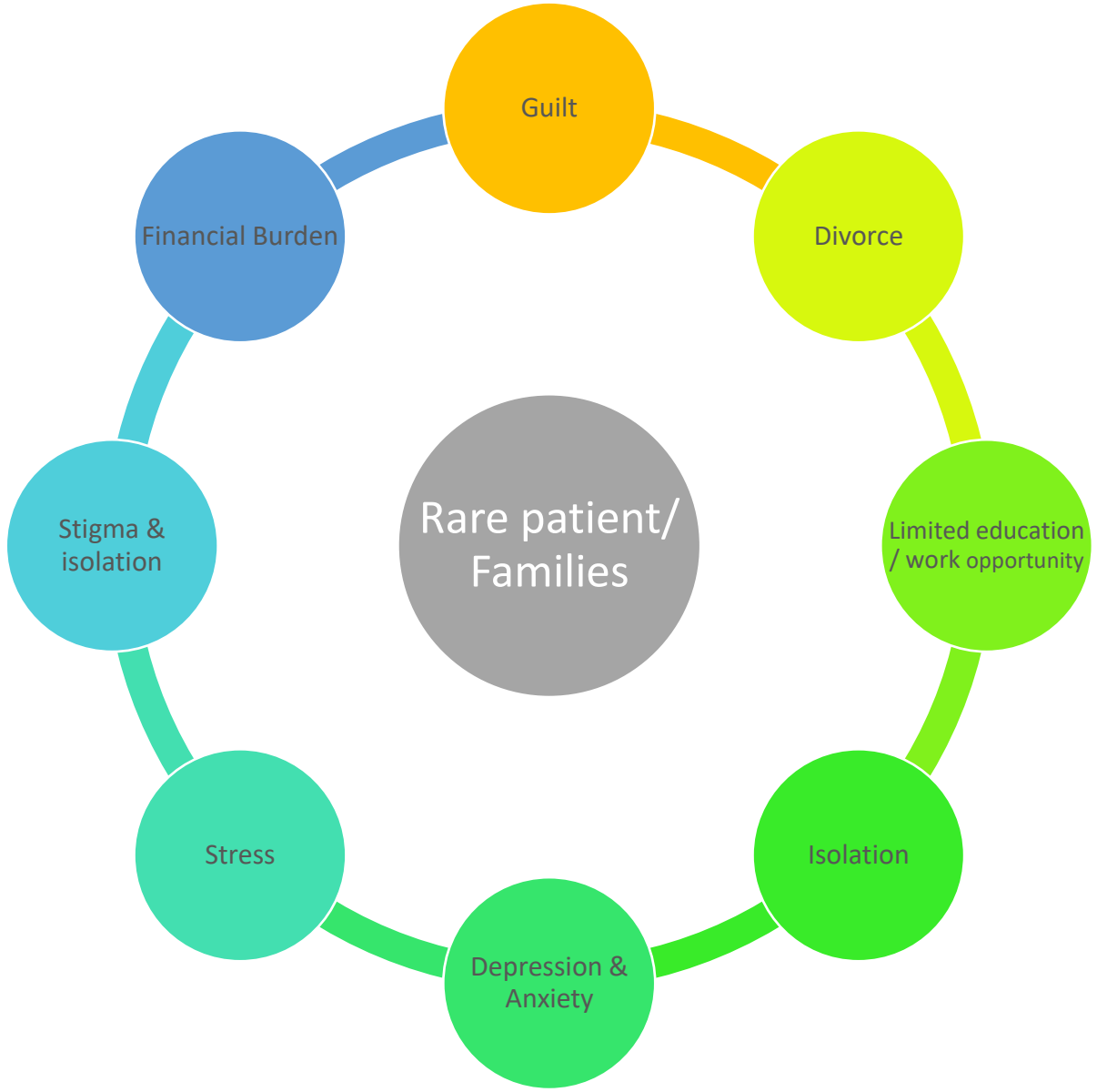
Solutions

Social Impact

The complex nature of rare diseases,

coupled with limited access to treatment and services,

means that family members are often the primary source of solidarity, support and care for their loved ones.



context to submissions

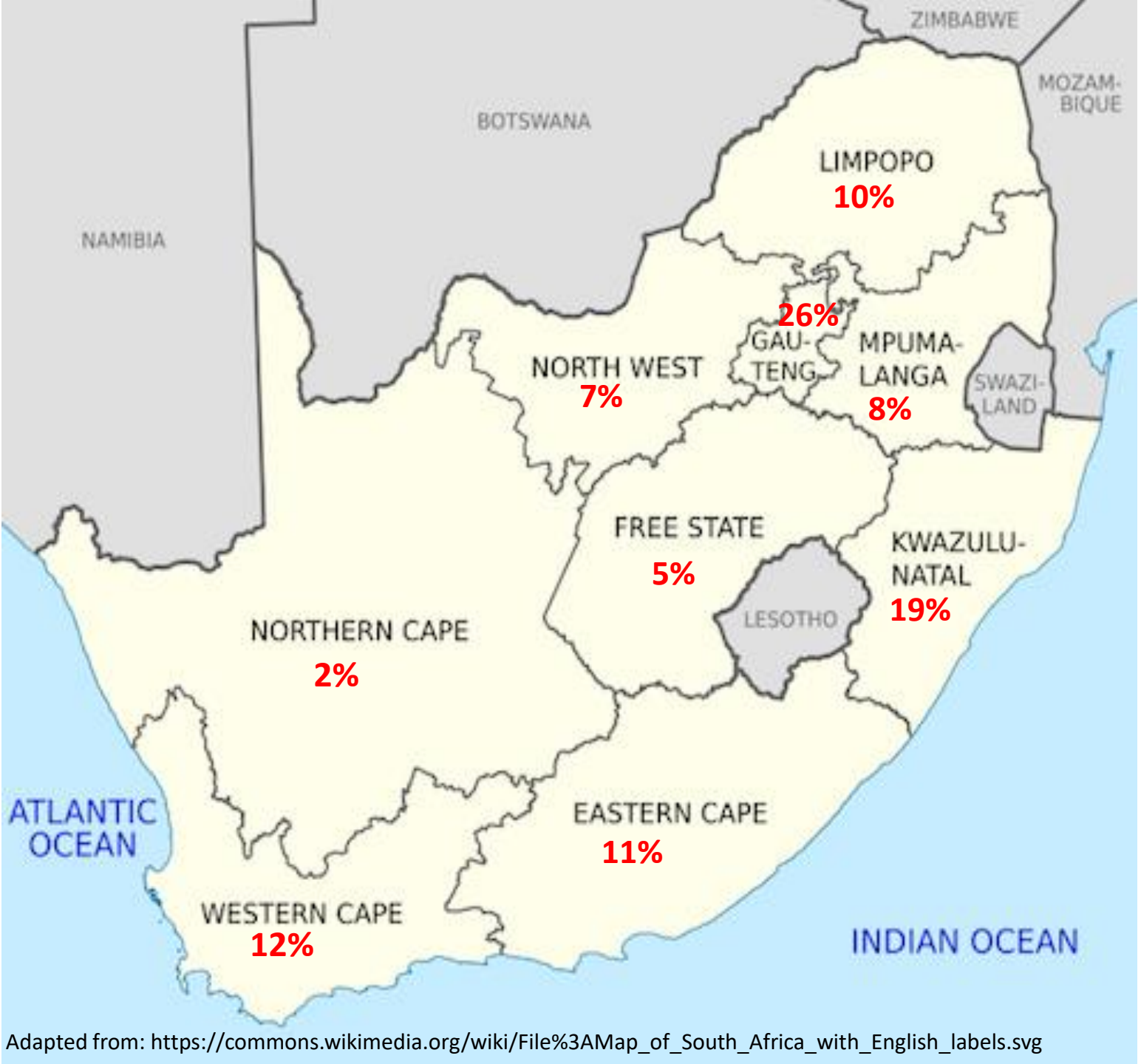
- RDSA is committed to the improvement of healthcare systems in South Africa and eager to find solutions to better meet the needs and ensure the care of the population of South Africans who are impacted by rare diseases and congenital disorders.
- RDSA has always been committed to engaging with the various stakeholders, including the National Department of Health to find solutions to access of healthcare for the rare disease and congenital disorder community.

2018 feedback

- Not clear where Rare diseases will receive funding.
- Clarity on how NHI will be funded.
- Prioritization of those most in need – need clarity on if rare diseases are included.
- Definition of quality health services is unclear.
- Discretion of the benefits advisory committee.
- Multi disciplinary, comprehensive and high-cost services may not be included.

2018 feedback

- Diagnostic Services – how will lack of current services and infrastructure be addressed.
- PMB Criteria – will existing diseases be removed / have less coverage?
- Progressive realisation – not providing treatment and services for rare diseases and congenital disorders is a violation of human rights.

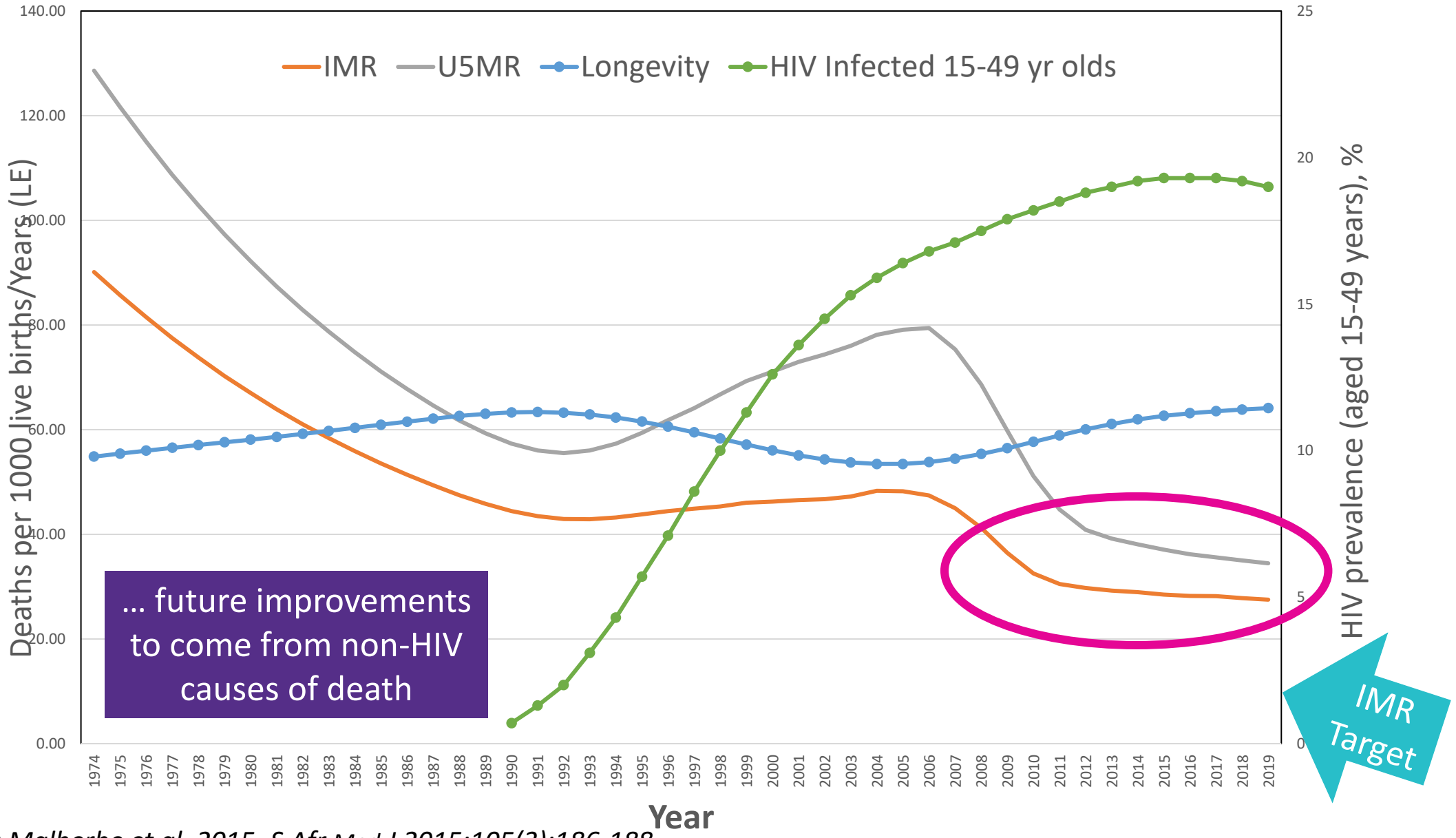


Adapted from: https://commons.wikimedia.org/wiki/File%3AMap_of_South_Africa_with_English_labels.svg

Profile

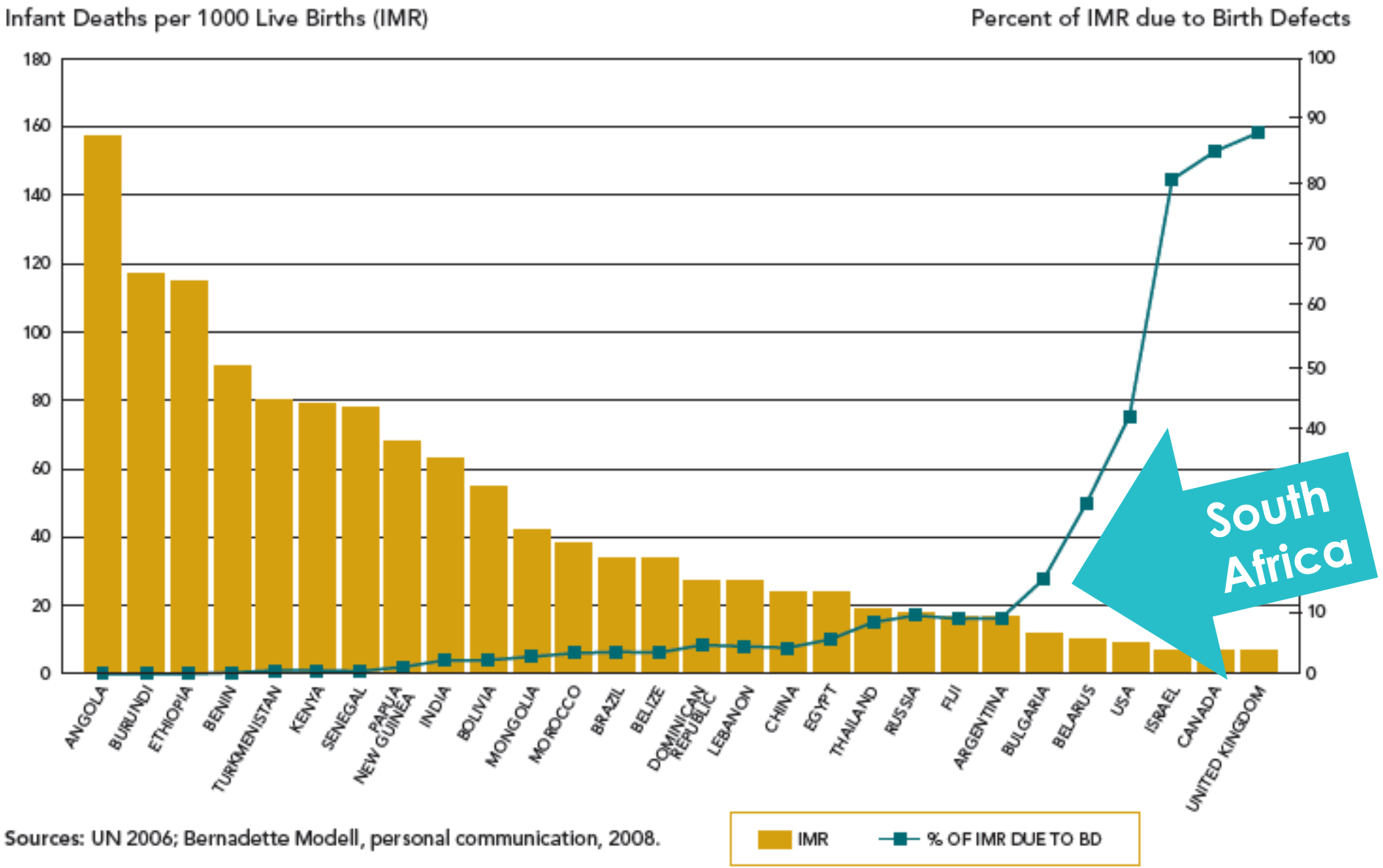
- **Population** 59,62m (2020)
- 29% < 15 yrs (2020)
- **Births**: 1,051,311 (2020)
- **Urbanised**: 63% (2012)
- **Fertility rate**: 1.9 – 2.9
- **LE** 62.5(m) & 68.5(f) (2020)
- **U5MR**: 34/1000 LB (2018)
- **IMR**: 25/1000 LB (2018)
- **NNMR**:11/1000 LB (2018)
- **HIV**: 13% of 15-49 yrs (2020)
- **AMA**: 16% (2019)
- 85% **state**:15% **private**

Epidemiological transition in South Africa 1974-2019



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

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.

Proportion of congenital anomaly deaths according to World Bank country classifications

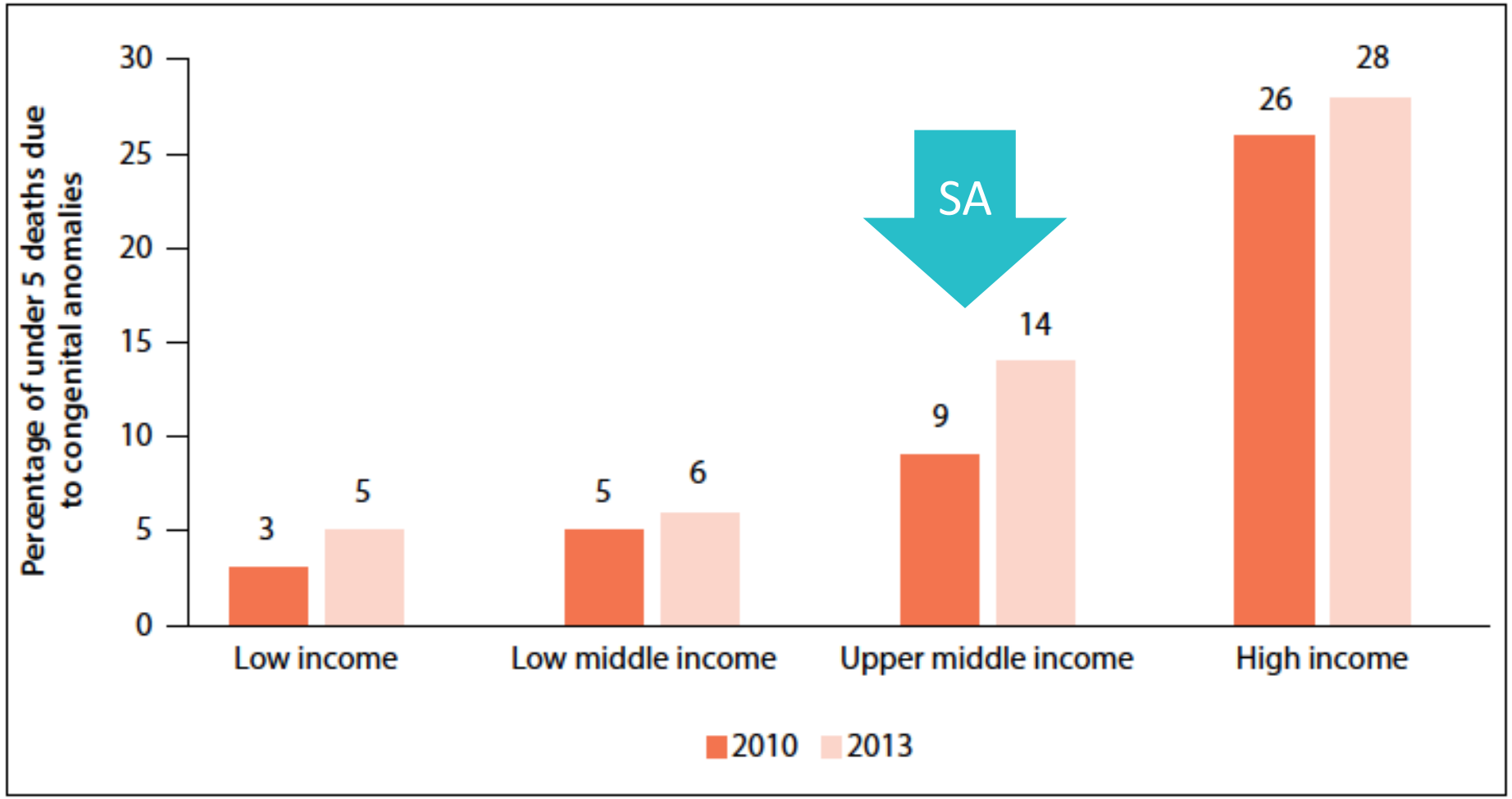


Fig. 1. Comparison of the percentage of under-five deaths resulting from congenital anomalies for World Bank Country Classifications. [9]

Malherbe et al, S Afr J BL, May 2016

2021 capacity

- Referral services particularly for genetic services
- Lack of human resources in medical genetics:

Table 1. A comparison of medical genetics services capacity in 2001 and 2021 in the state funded health sector

	Recommended* 2001	2001		2021		
Category	**Number/ratio**	**Number (Actual)**	**Ratio**	**Number (Actual)**	**Ratio (state patients)**	**Number Required**
Medical geneticists	**20/1 per 2m**	4	1 per 11.2 m	9	1 per 4.5 m	**25**
Genetic counsellors	**80/1 per 580 000**	<20	1 per 2.2 m	7.5	1 per 6.6 m	**85**
Medical scientists/technologists	**100/ 1 per 450 000**	50	1 per 900 000	TBC	TBC	**110**

Adapted from : (Malherbe et al, 2016)

Personal communications 28 July 2021: M. Urban, K. Fieggan, A. Krause, L. Yates, M. Conradie;

2021 provincial capacity

Table 2. Provincial breakdown of medical genetics services capacity in 2021 in the state funded health sector

	STATE FUNDED POSTS				
Province	**Medical Geneticist**	**Vacant Posts**	**Registrars**	**Genetic Counsellor**	**Vacant Posts**
EC	0	0	0	0	0
FS	0	**2**	0	0	0
GP	**4**	**3**	**5**	**5**	**1**
KZN	1	0	**2 vacant**	0	0
MP	0	0	0	0	0
NC	0	0	0	0	0
NW	0	0	0	0	0
WC	**4**	0	**2**	**2.5**	**0**
Total SA	**9**	**5**	**7**	**7.5**	**1**

Personal communications 28 July 2021: M. Urban, K. Fieggan, A. Krause, L. Yates, M. Conradie;

2018 feedback

- Congenital disorders and most rare diseases are a non-communicable disease (NCD).
- Represent significant burden of disease.
- Data deficit.
- Surveillance and monitoring underreporting by >90% (Lebese et al 2016)
- Collaboration with civil society.
- Essential Drug List/ access to appropriate medication.

2019 feedback

- Health system revolution through legislation.
- The inclusion of civil society and health service users in all decisions and processes.
- Governance under NHI.
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Conclusion

How can we claim to have achieved Universal Health Care, or health equity for all, when we knowingly exclude marginalized and vulnerable patients such as rare disease patients, ultimately leaving them behind?