The need for innovative thinking to treat Rare Diseases in Africa

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Rare Diseases

- Generally are chronically debilitating
- Referred to as Orphan Diseases due to the abandonment and lack of interest healthcare sector
- Collectively they are common - affect estimated 6-8% of the global population

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What are the Challenges?

- Treatment of RD's involve orphan products not commercially developed and very expensive.
- When treatment is available, often cost is the only barrier to accessing it.
- Healthcare regulations and policies do not recognize RDs as a threat
- Late diagnosis/lack of treatment may cause permanent disability and often death.
- Resources and Technology are allocated to common diseases in larger populations
- Africa is fighting major challenges in terms of non-communicable diseases which further increase the healthcare burden.

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What are the Challenges?

Inadequate level of care due to:

- Insufficient knowledge and information
- Poor diagnostic facilities
- Personal limitations related to socio-economic circumstances
- Inequality and inequity continues to exist in this community

Equality

Equality = SAMENESS

Essentially it means giving everyone the same thing

IT ONLY WORKS IF EVERYONE STARTS IN THE SAME PLACE

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Equity

Equity = FAIRNESS

Everyone has access to same opportunities

We must first ensure equity before we can enjoy equality.

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Reality

Reality = Inequality & Inequity

Those who shout the loudest generally get the most.

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The Goal: Liberation

The Barriers are removed, resulting in equality and equity equally.

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What is needed prior to developing a framework?

- Updated diagnostic codes that account for the majority of rare conditions – critical to determining actual threat and prevalence
- We need to define our definition of a rare disease. We cant be treating and advocating without clear boundaries and interpretation of who are subjects are and what their entry criteria is.
- We need to develop a plan regarding a registry – currently the only data available for rare diseases in SA is held by RDSA, and input is done by patients allowing for a high percentage of error and inaccuracies.

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What to think about in developing a framework to improve access.

- Rare diseases do not only require commercial therapies, supportive care and adjunctive therapies are critical to improve quality of life.
- Social considerations for patients & families are needed to be taken into account when considering access to treatment including: travel expense, need for compliance, level of care and infrastructure in rural areas etc.

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Social Considerations

- Rare diseases cannot be evaluated in isolation – as they do essentially affect the community as a whole.
- A rare diagnosis often leads to:

- Guilt/self-accusation
- Divorce
- Limited Work opportunity
- Social isolation/stigmatization
- Financial burden

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The ‘Un-curables’

- 95% of RD’s have no treatment/cure.
- Patients quality of life is not dependent on severity of the illness, but:
  - Availability of treatment
  - Health support services
  - Broader social support
  - Access to supportive therapies
- Most are not recognized by our healthcare system
- Lifespan is often reduced due to lack or loss of independence

(Rajmil et al. 2010)

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Research is necessary

- -Rare diseases are hard to study and time consuming
  - -Patient pool size limits clinical studies

    making post-approval studies vital.
  - Efficacy studies cannot be strengthened without collaborative effort.
  - Available financial resources to consolidate and publish data and facilitate research that is required.

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Research is necessary

Contents lists available at ScienceDirect

Molecular Genetics and Metabolism

journal homepage: www.elsevier.com/locate/ymgme

Alglucosidase alfa enzyme replacement therapy as a therapeutic approach for a patient presenting with a PRKAG2 mutation

Stephanie L. Austin [a], Andrew Chiou [a], Baodong Sun [a], Laura E. Case [b], Kenny Govendrageloo [c], Perrin Hansen [d], Priya S. Kishnani [a,*]

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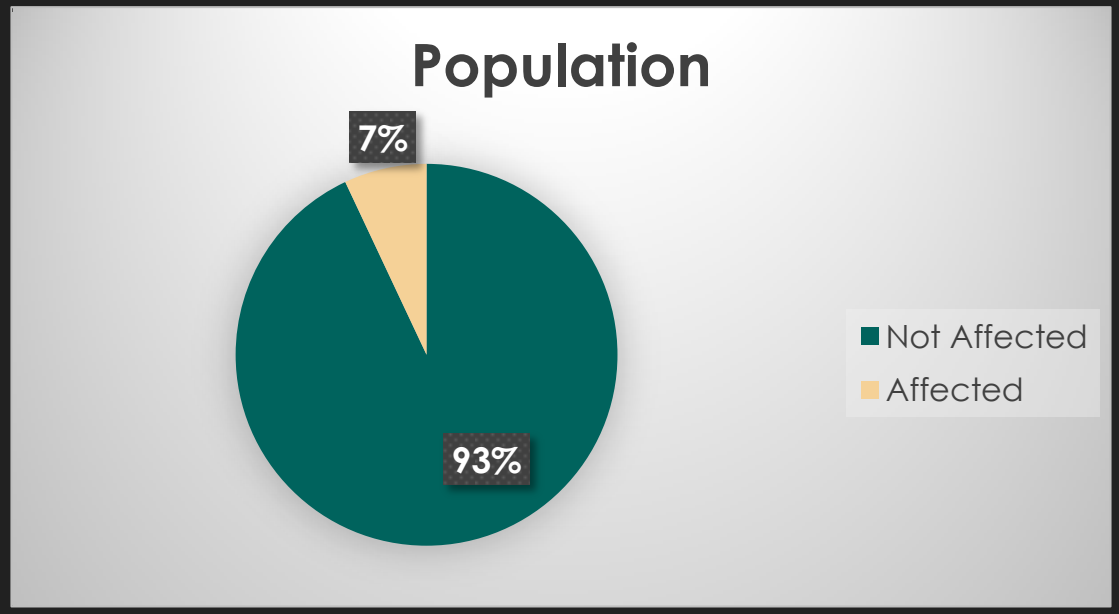
The BIG DEBATE – Can we afford to treat them?

On Average 7% of the population are affected by a RD



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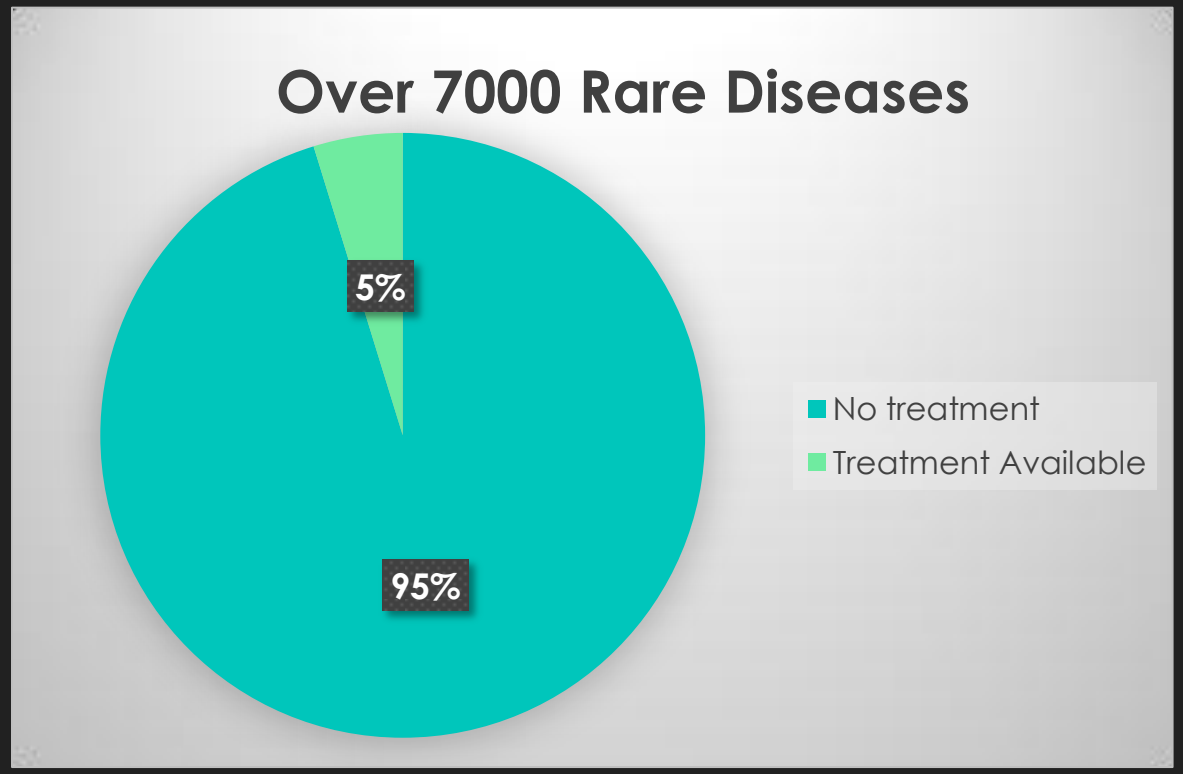
The Treatable Ones…

- Only 5% of all Rare Diseases are treatable



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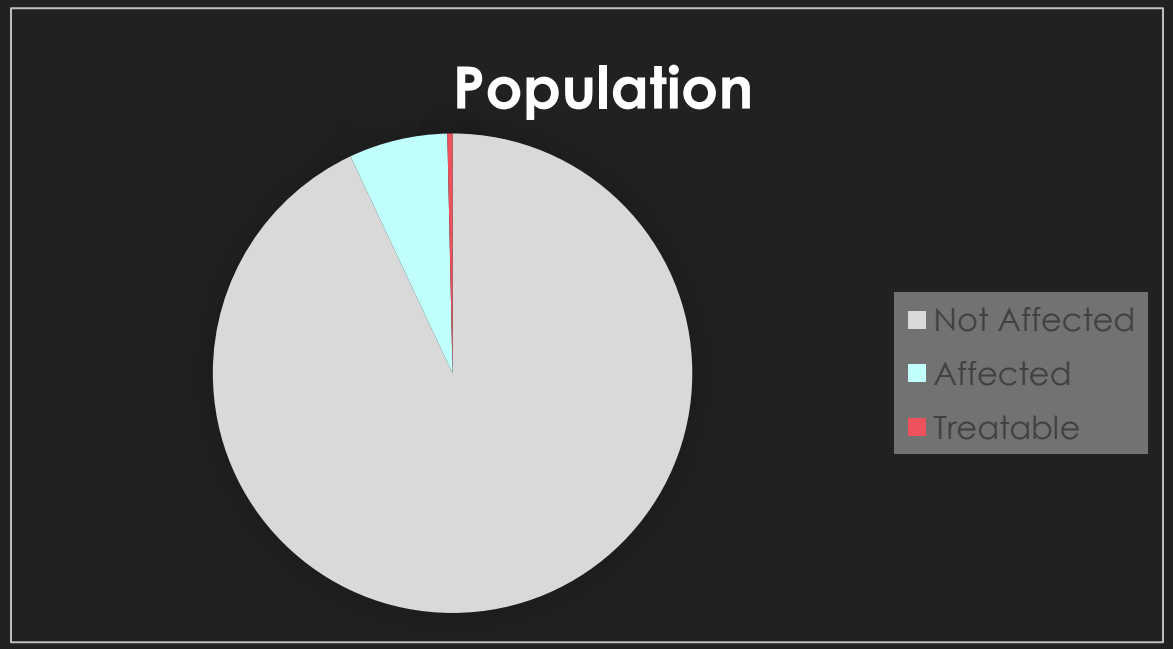
Combined…

0,35%…Is that really a Burden?



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Financial Burden

South African Case Study: LSDs with available treatment locally.

(MPS 1/ MPS 2, Pompe, Fabry and Gauchers)

- 112 Patients in total
- Listed as the most expensive top 20 drugs in the world
- Based on Europe Epidemiology, we should have ± 1700
- Average diagnosis rate of 7%

- In comparison to the **2013/14 State Health Budget**, To treat all of these patients would have used a total of 0,01%

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Possible Solution?

Risk equalization fund:
- International trend is to reserve 1% of the health budget to Orphan Products and specialized services
- Based on the 2013/14 figures,
1% = R2,7 billion
- A dedicated panel would form the advisory board

Include Rare Diseases onto existing policies – if it is good for Rare, it will generally be good for other disease groups too

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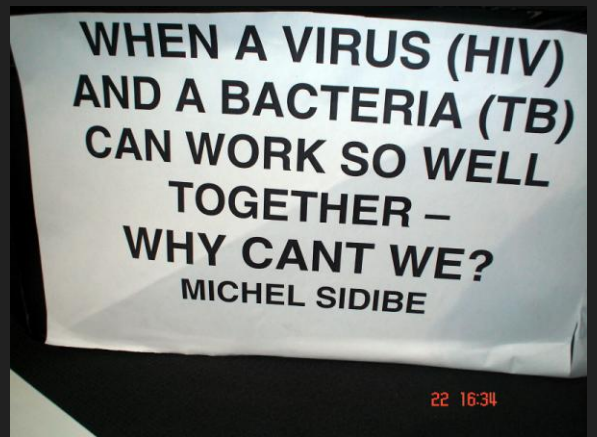

Some innovative thinking is required

- Smaller vial sizes for rare disease products
- Using complete vials
- Empowering patients to care better for themselves
- Home care
- Patient education
- Limiting retests etc

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Important things to remember

- NO disease is so rare not to deserve proper attention
- They are permanent, constantly progressing diseases and their development is unpredictable.
- Early identification and treatment lead to positive prognosis with improved quality of life.

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In Summary

- Untreated patients still place a burden on the healthcare system
  - Obligated to reduce the under 5 year mortality rate
    - Limited for choice in terms of treatment
- Yes, one RD patient costs equal that of 100 HIV babies, but how many of those patients are NOT being treated?
- Should funding be the decider between life and death ?

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