



RareX October 2016

Spier Conference Centre, Stellenbosch

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Vision:

To improve the lives of all those affected by congenital disorders in South Africa

- **Advocacy:** to promote accessible, effective and relevant genetic services for the care, prevention and treatment of CDs
- **Support:** Provide a network of support and fellowship for all those affected.
- **Education:** Promote the education of relevant medical health professionals.
- **Awareness:** Educate and raise awareness of the general public.
- **Research:** Facilitate, promote, and support research related to CDs in South Africa.
- **Sustainability:** Strive for long-term sustainability as an organization.
- **Growth:** Continue to grow and develop as an organization through continued learning at all levels.

Improving the lives of those affected by congenital disorders



Trisomy 18



Fragile X



Turner Syndrome



SA Thalassemia Association



Neurofibromatosis



Porphyria



RETINA SOUTH AFRICA
A Cure in Sight for Blindness



Internal

- Relaunch & buy-in
- Shift in mindset (charity to organisation)
- Sustainability (funding, leadership)

External

- Fragmentation/lack of unity
- Competing health priorities (communicable & other NCDs)
- **1 in 15 live births** (6.8%) affected in South Africa
- Medical genetic services declined (capacity) & health need increasing
- Lack of political commitment & funding
- Underreporting of CDs by over **98%** (Lebese et al, 2016)
- Lack of awareness & inadequate education
- Terminology confusion
- Attitudes: “nothing can be done” & over focus on prevention