5 year review
2014-2019

Rare
Diseases
South Africa

Patient Advocacy

Stakeholder Meeting – 27 Feb 2019
Premier Hotel, Johannesburg

Rare Diseases South Africa

RDSA undertook to host a meeting engaging with all stakeholders within the private sector of healthcare, to discuss the access challenges currently faced by members within the rare disease community

OBJECTIVES

- Gather and forge linkages between key, relevant stakeholders with a common interest in improving access to treatments for those impacted by rare diseases
- Provide a comprehensive overview of the rare disease access challenges within the healthcare sector
- Identify lessons learned and develop a way forward

Key Findings

- The need for an accepted definition of a rare disease
- The rigid regulations in terms of SEP pricing are inhibiting innovation in terms of market access
- Lengthily drug registration processes are inhibiting appropriate and timeous access
- Robust discussion and engagement on innovative access models are required
- The legal interpretation of existing law is variable
- Regulators need to ensure compliance of appropriate decisions on PMB legislation
- Exemption guidelines for evidence-based evaluation and drug registrations are needed
- Improved patient advocacy and patient participation is required in healthcare sector
- New-born screening is a possible cost-effective approach to early identification of treatable conditions
- There is disparity between access between different medical schemes as well as the state and private sector
- Further engagement is needed on a continuous basis

IMPACT

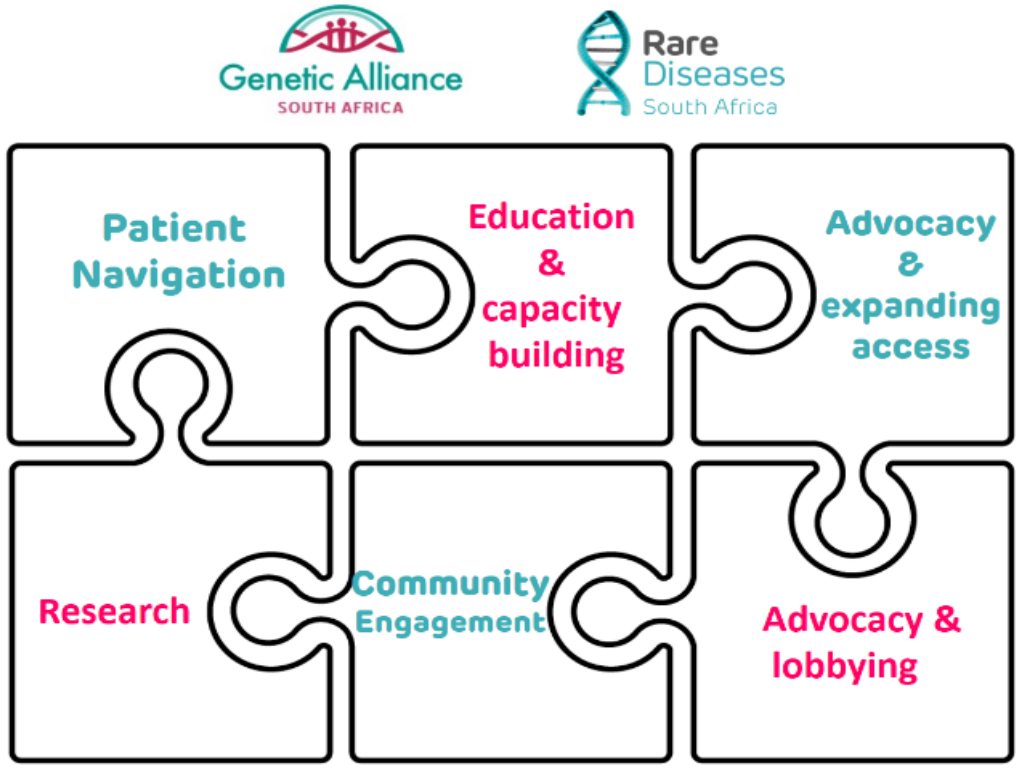
Private Sector multi stakeholder **Forum launched** to address Rare Disease challenges across the sector.

Alliance and Partnership Development

RDSA has entered into various collaborative alliances and partnerships with organizations aimed at improving the health care sector, and Rare Diseases across the globe.

Key Alliances and Partnerships

- Rare Diseases International
- Eurordis
- IRDiRC
- Africa-Rare.Org
- IAPO
- SAHTAC
- Genetic Alliance SA
- Childhood Cancer Network SA
- Palliative Care network (PATCH / ICPCN)
- Rare Diseases Africa Advisory Group



IMPACT

- Improved **collaborative efforts** across the health sector
- Improved **advocacy**
- Increased **capacity and visibility**
- International and regional **participation**

Legislative Input

Rare Diseases South Africa

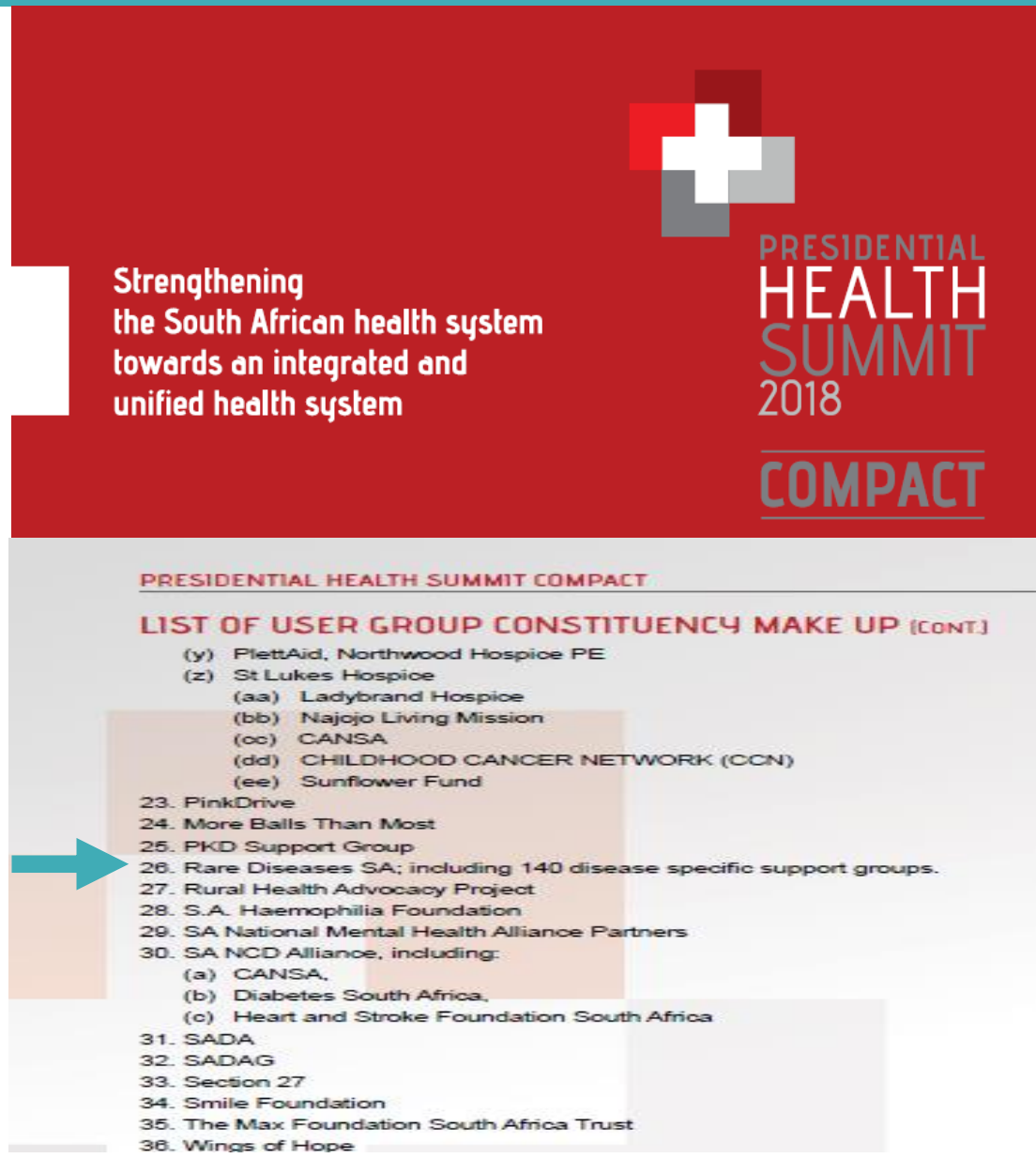
RDSA has provided input and feedback in various legislative and policy reviews across South Africa and internationally.

Legislative and policy input

- Presidential Health Compact
- PMB Review Process at Council for Medical Schemes
- SAHPRA Guidelines for section 21 access
- Genetic Guidelines drafted at NDOH
- NHI Bill
- Medical Schemes Amendment Bill
- ASSAF consensus on genomics
- Chronic Illness in Schools education policy
- Universal Health Care at WHO / United Nations

IMPACT

- Improved **collaborative** efforts across the health sector
- Improved **advocacy**
- Increased **capacity and visibility**
- International and regional **participation**
- **United** patient voice



Conference Participation

Rare Diseases South Africa

Kelly du Plessis, CEO of Rare Diseases has presented on various subjects at a variety of international and national conferences.

Role of patient organizations - University of Pretoria Metabolic Symposium, Pretoria, South Africa, 2013
Where we have come from and where we are going - Gaucher Symposium, Johannesburg, South Africa, 2014
The Big Debate: ethically acceptable or unacceptable social consequences - Paediatric Refresher Course, Cape Town, 2016
Re-inventing the future for Rare Disease patients – Pompe Symposium, Johannesburg, South Africa, 2015
Rare diseases from a parent's perspective - ICORD, Mexico City, Mexico, 2015
The need for innovative thinking to treat Rare Diseases in Africa – SAPHEX, Johannesburg, South Africa, 2016
Private Practice: The good, the bad and the Ugly – UpToSPAED, Johannesburg,South Africa, 2017
The Need for collaboration in the Rare Disease environment – Pfizer Symposium, Johannesburg, South Africa, 2017
Challenges of Rare Diseases in developing countries – Rare Diseases International, Madrid, Spain, 2017
The need for long term sustainable solutions - Sanofi Scientific Forum – Magaliesburg, South Africa, 2017
The Status Quo of Rare Diseases – World Orphan Drug Congress, Washington, USA, 2016
Keynote Address: Palliative Care in Rare Diseases: are we swimming upstream? ICPCN Conference, Durban, South Africa, 2018
The Long term impact of Rare Diseases - Fabry Symposium – Shire, Johannesburg / Durban / Cape Town, South Africa, 2018
Keynote Address: The Multidisciplinary Approach – RareX, Johannesburg, South Africa, 2018
United Nations: Rare Diseases in South Africa – New York, USA, 2019

IMPACT

- Improved **collaborative** efforts across the health sector
- **Education** of HCPs and Stakeholders on the plight of Rare Disease patients
- Increased **funding**
- International and regional **participation**
- **Sharing** of best practice and expertise

Education

RareX Bi-Annual Conference (2016 - Cape Town / 2018 - Johannesburg)

Rare Diseases South Africa

Theme: The need for a multidisciplinary approach

Delegates

- 2016 182 Attendees
- 2018: 125 Attendees
- 2016: 16 Support Groups
- 2018: 19 Support Groups

Sessions

- Genetic Landscape in South Africa
- Inborn Errors of Metabolism
- Palliative Care in South Africa
- Diagnostic availability
- Global rare disease policies and programmes
- Access to treatment
- Research and Funding
- Improved quality of life approaches
- Patient Organizations
- Capacity Building with Rare Diseases Int.

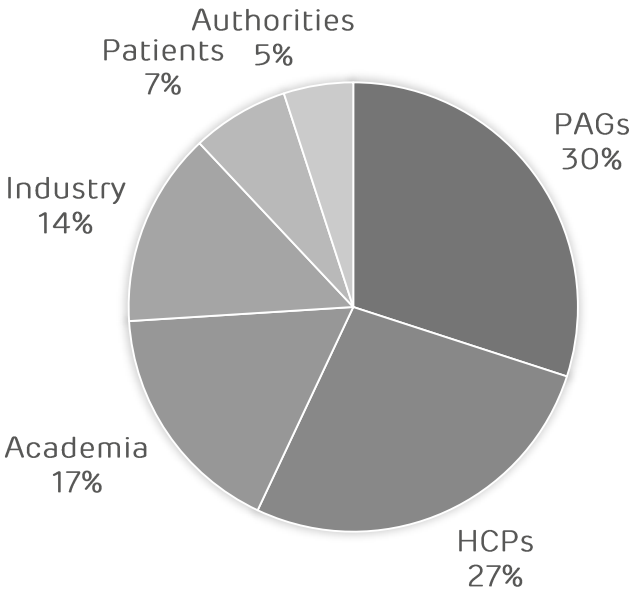
Post Event Feedback

- 48 Submitted Abstracts
- 89% of respondents said the technical information gained was good.
- 93% said the practical information gained was great
- 90% said new ideas were gained
- 82% enjoyed the timing of the discussion periods

"I took home a tremendous amount of knowledge, which was worth the travel and time. I think that many more conferences could benefit from all the diverse, participatory and collaborative approach which was adopted during this event."

RareX 2016 Delegate

DELEGATES



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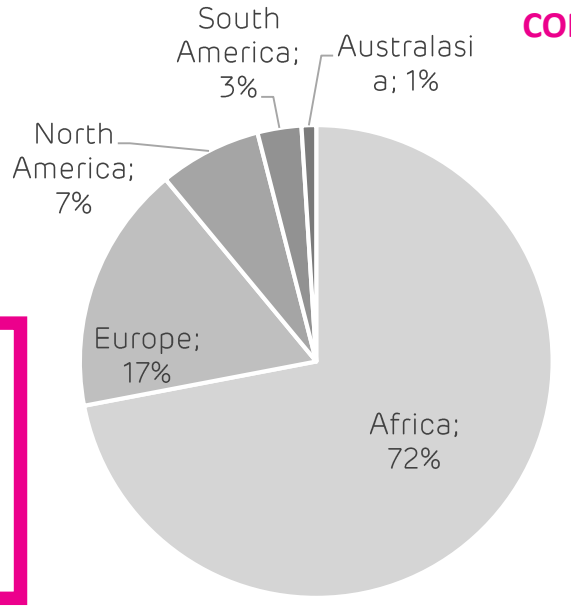


IMPACT

- **Created** patient centric approach
- **Educated** patients and caregivers, HCPs and policy makers
- **Sharing** of best practices
- **Connected** the community

SOS Feeding Course – 21-24 August, The Capital Hotel, Sandton

Rare Diseases South Africa

In partnership with Sandton Feeding Centre

The Sequential Oral Sensory (S.O.S) Approach to Feeding conference is a trans-disciplinary program for assessing and treating children with feeding difficulties and weight/growth problems from birth to 18 years. It integrates posture, sensory, motor, behavioural/learning, medical, and nutritional factors to comprehensively evaluate and manage children with feeding/growth problems.

Speakers:

- **Kay A. Toomey, PhD** - Pediatric Psychologist; President of Toomey & Associates, Inc.; Clinical Director of SOS Feeding Solutions at STAR Institute for Sensory Processing Disorder
- **Erin S. Ross, PhD, CCC-SLP** - Neonatal and Pediatric Speech Pathologist; Assistant Clinical Professor at University of Colorado; Faculty at Rocky Mountain University of Health Professions; President, Feeding FUNdamentals, LLC

Delegates: 92 **CPD Points:** 32 points

Course Objectives

- Identify oral, sensory, motor, cognitive and emotional developmental milestones key to feeding.
- Recognize and describe the major reasons why children won't eat, as based on learning theory principles.
- Identify physical, behavioural, motor, oral-motor, and sensory factors as a part of a Feeding Assessment.
- Apply behavioural and social learning principles, and systematic desensitization to feeding problems.
- Create and implement a SOS feeding program for babies, toddlers and young children, in group and individual treatment formats.

IMPACT

- Improved **knowledge** base for HCPs working with rare disease patients who are fussy feeders and not able to gain optimum nutrition
- **Improved** patient centric approach

Pilot Port Training in partnership with B Braun Medical

Rare Diseases South Africa

RDSA, in partnership with B Braun Medical developed a pilot port access training program for patients, caregivers and nursing professionals.

OBJECTIVES

- Build awareness on ports and
- Educate delegates on the correct ways of using a port
- Connect and educate our key influencers (doctors/nurses) that influence the patients end decision.

Key speakers included:

- Kelly Du Plessis (CEO RDSA)
- Paul Gardiner (Past CEO of Discovery Homecare)
- Phindile Llale (B Braun representative)
- Sr. Andile Zwane (Registered Nurse)

How many were trained: 20 delegates
Who was trained: Patients, nurses, HCPs

IMPACT

- **Provided** responsible, patient-centred approach to long-term IV access
- **Reduced** risk of infection for users
- **Improved** nurses / HCP ability to access ports

KEY LEARNINGS PRIOR TO WORKSHOP

- 70% of Rare Disease patients that require long-term IV treatment, of which 42,8% do not know what a portocath is.
- ½ the patients using a port have had an infection
- Almost all patients using a port have not had formal training on how to use a port

#WebinarWednesdays

Rare Diseases South Africa

GOAL: A monthly online education platform aimed at educating those within our community around a variety of topics.

Topic	Speakers	Objectives
Compassion Fatigue	**Kim Ballantine:** Professional Business and Life Coach, Industrial Psychologist, Motivational Speaker @ Camber Coaching	• Increase self awareness around compassion fatigue and associated symptoms • Provide coping skills to caregivers impacted by burnout
Navigating Healthcare Benefits (Launch of Rare Assist)	**Jerome Pillay:** Strategic Risk Assessor @ Rare Assist **Aneesa Ally:** Strategic Risk Assessor @ Rare Assist	• Improve patient understanding on reimbursement process at Schemes • Provide platform for patients to escalate claims • Improve knowledge around PMBs and Formularies
Finding Your Voice	**Claudia Ferguson:** Business Director @ Orange Ink PR **Megan Hunter:** Rare Disease Ambassador **Roxy Burger:** TV Personality and Celebrity Blogger	• Assist patients and champions in developing key messaging for media • Encourage patients to find their voice and share their story • Ensure patients are well placed to engage media on impact of rare diseases
Ehlers-Danlos Syndrome	**Lara Bloom:** Executive Director, International @The Ehlers-Danlos Society **Dr Alan Hakim:** Rheumatologist, United Kingdom	• hEDS Patient Registry education • Improve understanding on supportive care for EDS • Understanding the genetics around EDS
Terminal Illness? What to consider...	**Dr Mehnaaz Akbar Ally:** Paediatric Palliative Care Practitioner @WitsPal & Lambano Sanctuary **Adv Bonnie Venter:** Legal Affairs @Transplant Education for Living Legacies (TELL) **Petra Burger:** Social worker, Palliative Care Practitioner, Social Entrepreneur, Trainer and Facilitator and Disability Management Specialist	• Educate patients on palliative care and when it should be implemented • Dispel the myths around end of life care • Educate patients and families on Organ Donation Consent • Educate patents and families regarding living wills and legality of "D.N.R forms" • Facilitate referrals for patients and families to palliative care services

IMPACT

- Improved patient and community education
- Increased support for those impacted by Rare Diseases
- Improve referral pathways

Community Engagement

Acromegaly & Cushings patient day 2017

Rare Diseases South Africa

First ever Acromegaly & Cushings Patient Day, proudly sponsored by Novartis.

This event brought together 22 people across SA – of which 6 were Acromegaly patients and were 3 Cushings patients - for an informative, supportive, patient focused meeting.

The speaker line up included talks from Genetic Counsellor, Sarah Walters, CEO of Rare Diseases South Africa, Kelly du Plessis and Academic Researcher, Dr Helen Malherbe from Genetic Alliance, who covered topics such as genetic counselling and the importance thereof, advocating for patients with rare diseases and the benefits of a support group.

IMPACT

- **Share** organisational and advocacy knowledge in terms of access to treatment.
- Provide information on the diseases and **treatment** outcomes
- Provide **motivation** and support those newly diagnosed.
- Raise **awareness**
- Provide a platform for patients and advocates to **interact** and role-play when communicating their story.
- Reduced **stigma**

Patient family day 2018

Rare Diseases South Africa

RDSA has realised the value of community and often aims to provide an opportunity for our patients and their families to meet and connect with others in our rare disease community.

Where:	Tembali Party Park, Chartwell, Johannesburg
Who attended:	108 patients and family members attended this occasion
MC:	Nicole Capper, Rare Diseases Ambassador and WarriorMom

"Alone, we can do so little; together, we can do so much"

~ Helen Keller

"OPEN ME WHEN" letter initiative – Mandela day 2018

In celebration of Mandela Day we encouraged our members to participate in our "Open me when" letter initiative, which will give love, encouragement and support to our vulnerable community when most needed.

The concept is simple – write a letter or create a small care package for any 6 or 7 of the listed 'situations' which rare disease patients may encounter and for which your letter and words of strength and motivation will make an enormous difference. Rare Diseases South Africa will provide the name, age and short biography on the patients for whom your letters/packages will be sent.

The letters need to be addressed for the following 'open me when' occasions:

Open when needing motivation
Open when you need to smile
Open when you're having a great day
Open on your birthday
Open when you are scared
Open when you are in hospital
Open when you are hungry

IMPACT

- 112 Patients emotionally **supported**
- Created **awareness**
- Allowed members of public to **"get involved"**

Rare Diamond Awards - 2017

Rare Diseases South Africa

Goal: Recognise excellence in advancement of overall healthcare, treatment and support offered by all those working within the Rare Diseases space in South Africa as well as individuals that have made a positive impact on the rare disease community.

Award Attendees: 138

Nominations Received: 77

Rare Diamond Winners

Rare Diamond in Advocacy – Dr Helen Malherbe honouring her commitment to ensure our patients' needs remain top priority and that congenital disorders remain top of mind for policy-makers.

Rare Diamond in Healthcare – Dr Fiona Kritzinger for advancing patient centred care transforming health care in South Africa.

Rare Diamond Project – Iris House for who have led and implemented innovative projects that deliver better health care solutions in the paediatric hospice space.

Rare Diamond Support Group: CureSMA for their commitment to support and work closely with people affected by Spinal Muscular Atrophy.

Rare Diamond Tribute – Melissa Platt, on behalf of Samuel Frederick Platt whose legacy continues to change the way palliative care is seen and offered to those at end of life.

Rare Warrior – Ashleigh Paterson honouring her life as a rare patient and her positive attitude and ability to live beyond her diagnosis.

IMPACT

- Developed **Champions** for Rare Diseases SA
- Increased **awareness** and created a public profile for those positively impacting our community
- Benefit from a better sense of **community**

Denim Walk 2019

Rare Diseases South Africa

Rare Diseases South Africa (RDSA), in conjunction with the Genetic Alliance South Africa (GA-SA), supported those affected by rare diseases or at risk of genetic disorders by hosting a Denim Walk.

Goal: Aimed at increasing the visibility of rare diseases and congenital disorders among the public and connecting the community, patients, their families, and support groups, to raise funds and awareness for both causes.

Route Distances: 1.5km (wheelchair friendly), 2km; 2.5km
Participants: Adults: 166 Visitors – Children: 32 Market Vendors:15
Sponsors: Novartis, MixFM (Broadcast live from the gardens / interviewed RDSA CEO, Kelly du Plessis and RDSA 2019 Brand Champion, Roxy Burger)

IMPACT

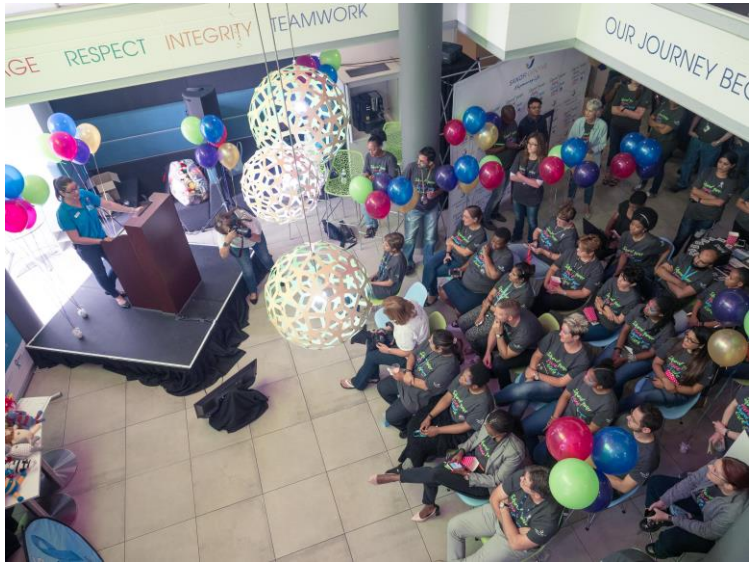
- Increased public **awareness**
- Increased **brand** awareness
- Raised **Funds** – R10 030,70 (after expenses)
- Allowed members of public to **"get involved"**

Rare Disease Day 2014-2019

Rare Diseases South Africa

RDSA is the official host of Rare Disease Day in South Africa. Taking place on the last day of February every year, this is the one day when all patients and families stand together united to educate those around us about our plight.

The denim ribbon is a perfect universal sign for rare and genetic disease awareness: Both are universal, come in pairs and are unique to the individual. In addition, blue is the colour of strength and the blue jeans denim ribbon is an ideal symbol that expresses solidarity and a simple concept that anyone can embrace to raise awareness and funds for rare and genetic disorders.



IMPACT

- Increased public **awareness**
- Increased **brand** awareness
- Raised **Funds** – R350 000 in 5 years
- Allowed members of public to **"get involved"**

#ShowYourRare

Capacity Building

Generation Rare - Support group incubation

Rare Diseases South Africa

Incubate (v.) grow under conditions that promote development.

Goal: taking nascent and budding support groups, and generating them into being fully fledged, independent patient advocacy groups able to better support and advocate for patients impacted by their focus area.

Inputs for critical success:

- Furnished office space including desks and access to boardroom facilities
- IT infrastructure & support: Wi-Fi, printers and phones
- Linkages with reputable suppliers
- Registration with all formal charitable entities (SARS / CIPC / Department of Social Development)
- Legal & financial support for compliance issues
- Fundraising mechanisms
- Staff to assist with mentorship

SA support groups that have been supported

- Prader-Willi Association of SA
- Sickle Cell Support SA
- Olive Children's Foundation
- Pulmonary Hypertension Association of SA
- Lysosomal Storage Disorders SA
- CureSMA
- FOP SA
- EDS Support SA

International Groups

- Rare Diseases Kenya
- Rare Diseases Botswana
- Rare Diseases Ghana
- Rare Diseases Zimbabwe
- Okanti Foundation (Namibia)

IMPACT

- Strengthened patient **advocacy** groups
- **United** voice across the health sector
- **Collaborative** input on advocacy efforts
- **Sustainable** emotional support for patients

Patient Group Huddle – 2017/2018

Rare Diseases South Africa

In partnership with Genetic Alliance SA and Campaigning for Cancer.

Goal: Bring together local and regional patient advocacy leaders and nascent patient groups and individual patients and caregiver advocates to assist with:

- Share organizational and advocacy knowledge and skill as part of on-going capacity building aims of more established or umbrella patient groups.
- Connect nascent groups and new patient advocates with resources to assist in their development.
- Provide peer exchange and input from subject matter experts.
- Provide a platform for patients and advocates to interact and role-play when communicating their story.
- To provide motivation and support those newly exploring advocacy.

2017 Huddle
Attendees: 98 patient advocates
Venue: Premier Hotel – OR Tambo
African Countries Represented: 8

2018 Huddle
Attendees: 56 patient advocates
Venue: RareX 2018
African Countries Represented: 14

IMPACT

- Strengthening of patient advocacy groups across Africa
- Empowered patient advocates with improved knowledge
- Increased capacity across the healthcare sector

Awareness

Brand Champions

Rare Diseases South Africa

Goal: Identify members of the Rare Disease community who are well placed to build awareness and represent RDSA in the media.

Oscar von Memerty

Oscar is a MPS VI patient and went around the country with RDSA in 2015, educating schools and corporates on Rare diseases.

Focus: Stigma and bullying associated with Rare Diseases

Nicole Capper

Nicole is a #MedicalMomma to daughter, Tatum , who was diagnosed with Cystic Fibrosis. Nicole has gone on to become Mrs South Africa, as well as being a contestant on SurvivorSA as our brand ambassador.

Focus: Addressing the challenges around invisible illnesses.

Roxy Burger

Roxy Burger is a #WarriorMom to daughter, Adrienne, who was diagnosed Congenital Hyperthyroidism at birth after newborn-screening being performed.

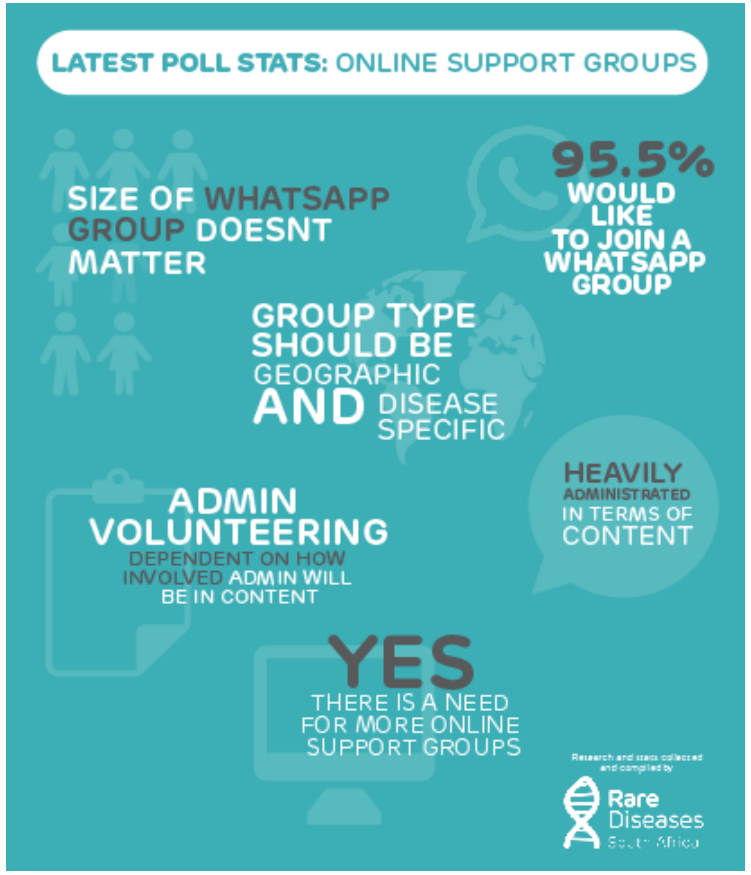
Focus: Benefits of Newborn screening as well as that Rare diseases can be treated with inexpensive, effective treatments.

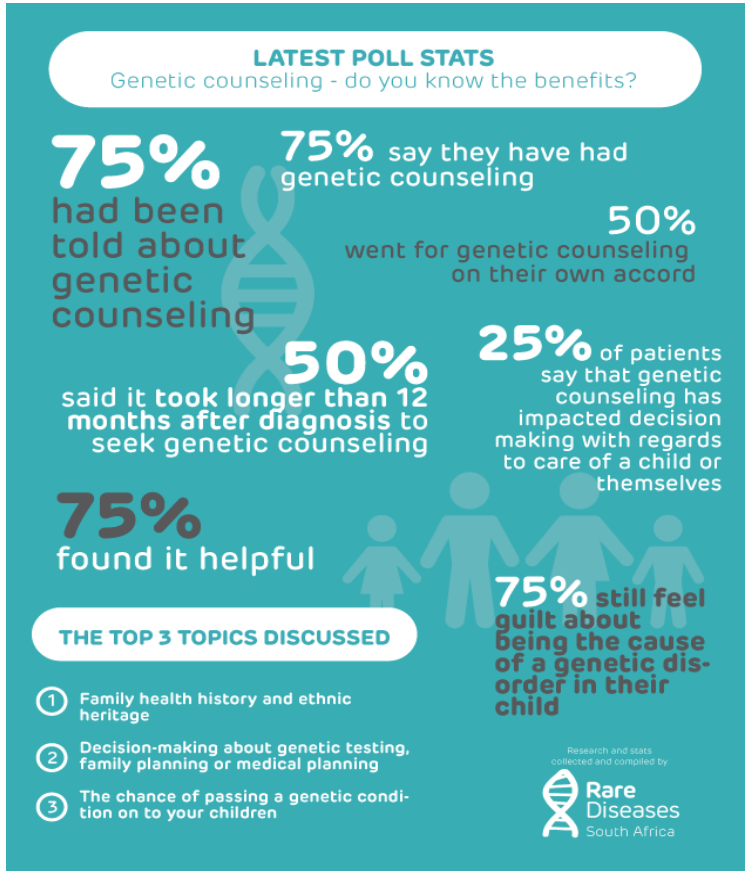
IMPACT

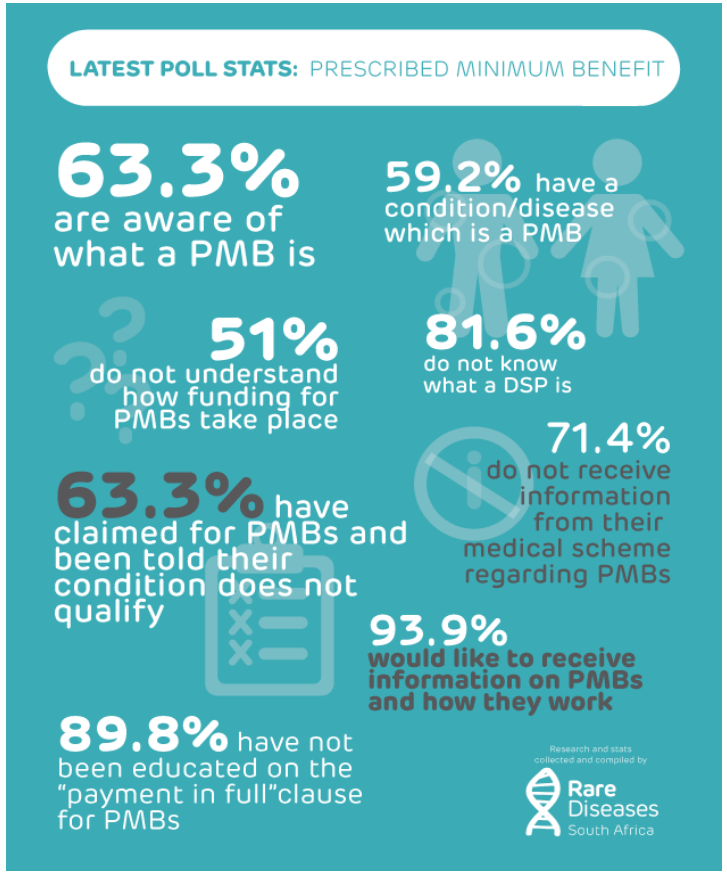
- Raising **awareness** on Rare Diseases and pertinent challenges on a peer-to-peer scale
- Brand Awareness and **visibility**
- Encourage **critical mass** and buy-in from general members of the public

Surveys and Polls

Goal: Identify and understand challenges and pathways impacting the Rare Disease community







IMPACT

- Improved **understanding** and knowledge base
- Increased **patient –centricity**
- Sharing of **best practice**
- Improved **patient navigation**

Disease specific awareness days

Rare Diseases South Africa

Goal: Promote knowledge and awareness on a specific disease within the public at large.

IMPACT

- Improved Peer-to-peer **support**
- Sharing of **best practice**
- Improved **patient navigation**
- Increased **awareness**

Disease specific ambassadors

Rare Diseases South Africa

Goal: Promote knowledge and awareness on a specific disease on a peer-to-peer platform.

RDSA knows that there is no better expert on a rare condition than a patient themselves which is why we make ambassadors of those who are energetic, passionate want to be patient experts who then lead the community.

These ambassadors are tasked with:

- Engaging with community on peer-to-peer level
- Identifying unique challenges experienced by the community and raising this with RDSA
- Providing responsible information and education to patients
- Monitoring communication platforms
- Identifying patients in need of further referrals
- Representing RDSA at disease specific meetings and conferences
- Gathering consensus on general advocacy activities for inclusion

IMPACT

- Improved Peer-to-peer **support**
- Sharing of **best practice**
- Improved patient **navigation**
- Increased **awareness**

Current Audiences

Rare Diseases South Africa

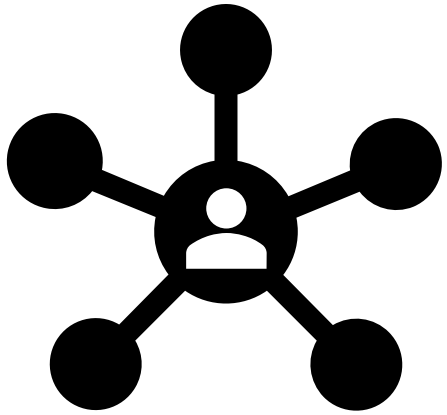
3500+ LinkedIn connections

8000+ newsletter subscribers

1500+ patients on database

2700 Instagram followers

Rare Diseases SA Community



650+ diseases

300 Volunteers

80+ Whatsapp patient connect points

46000+ Facebook followers

1500+ Rare ACTIVists

1580 healthcare professionals

4500+ Twitter followers

700+ Caregivers

140 Disease specific support groups diseases

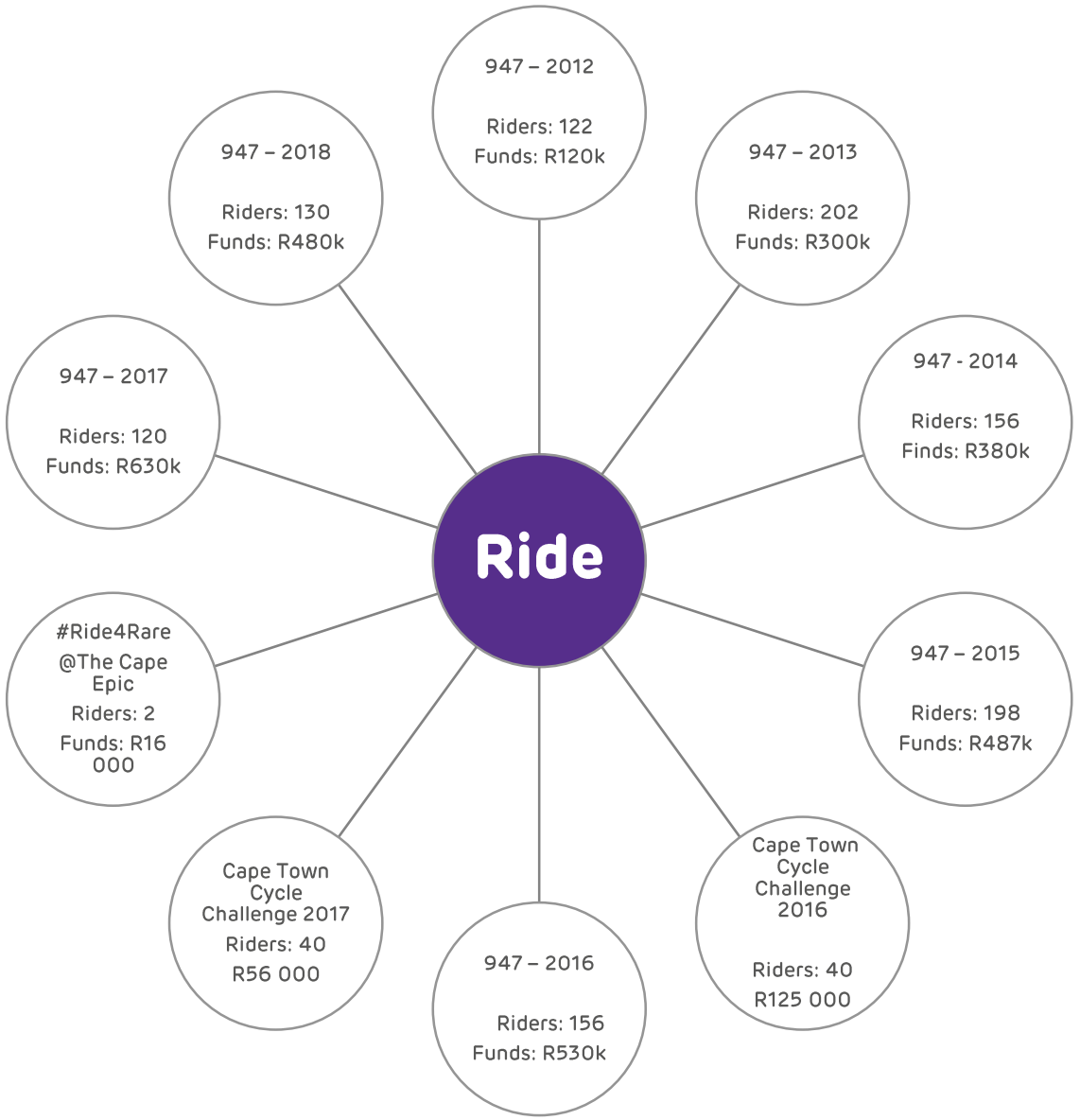
63 Medical Schemes

R.A.R.E ACTIVists

(R)aising
(A)wareness through
(R)ecreational
(E)vents

#Ride4Rare

Rare Diseases South Africa

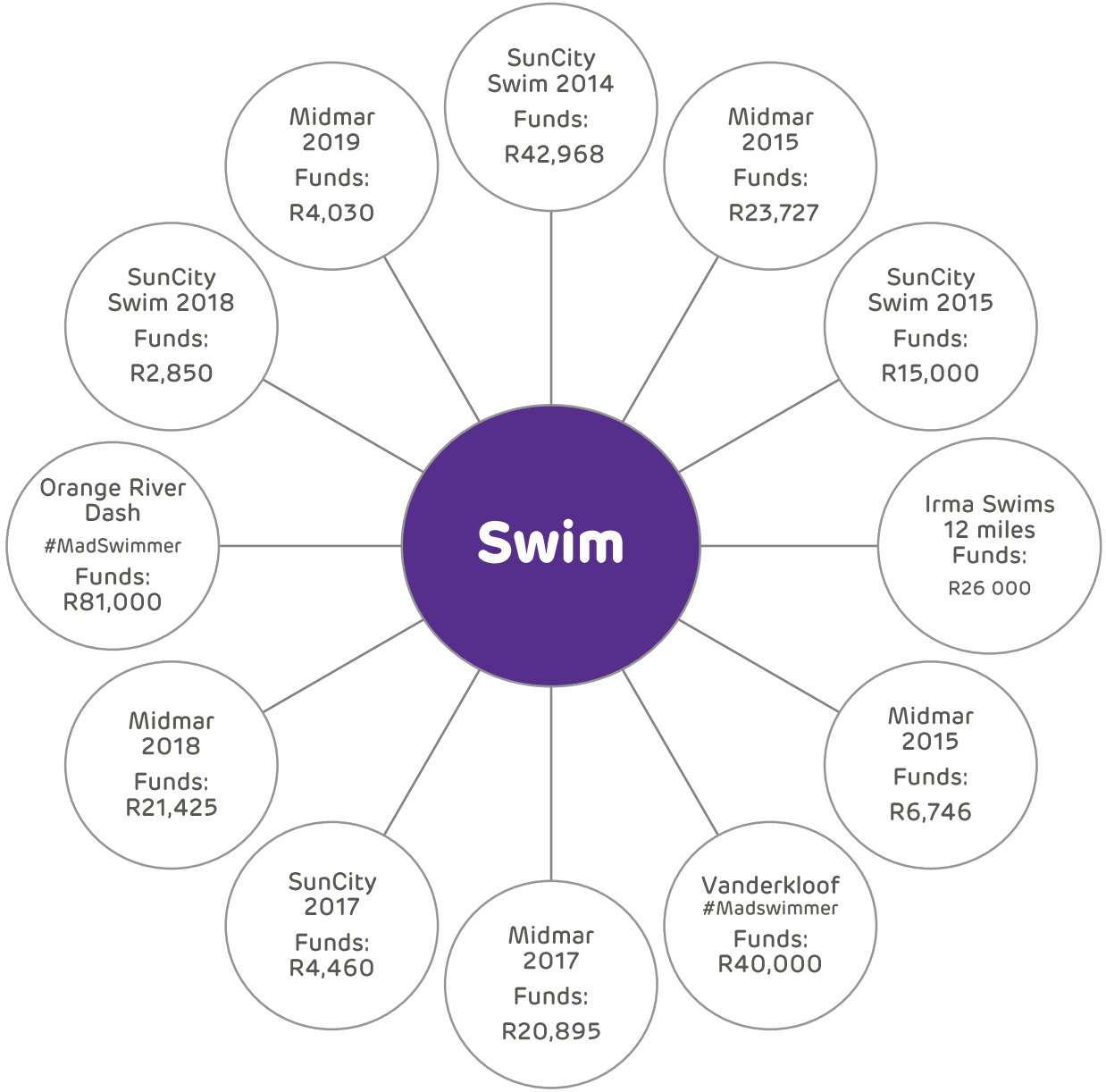


IMPACT

Number of Riders: 1166
Total Amount Raised: R3 124 000
Awareness: 162 000 people
1 patient positively identified and started treatment as result of our team.

#Swim4Sadie (in memory of Sadie Penn)

Rare Diseases South Africa

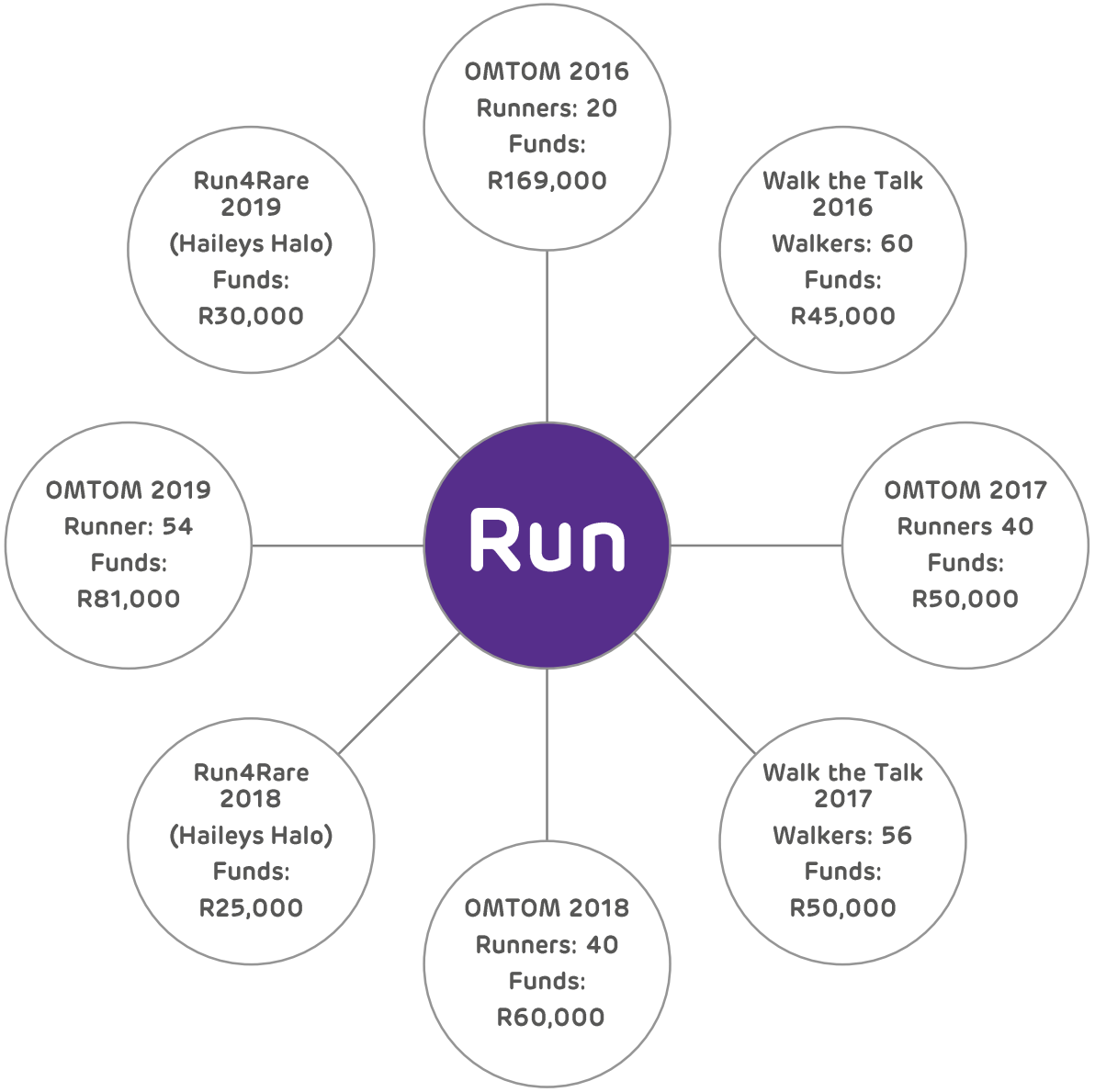


IMPACT

Number of Swimmers: 64
Total Amount Raised: R289 101
Awareness: 9 750 people

#Run4Rare

Rare Diseases South Africa

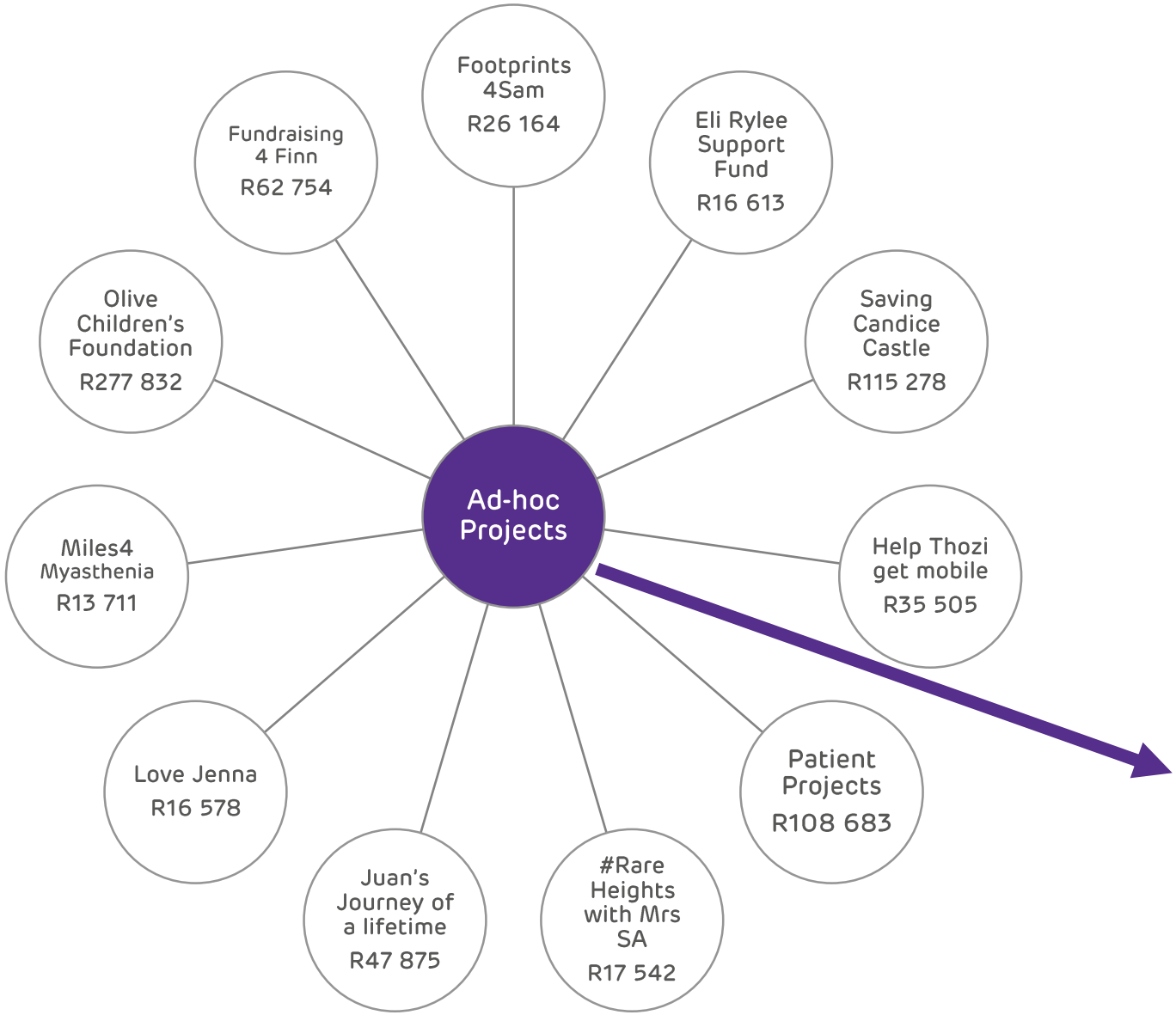


IMPACT

Number of Runners: 270
Total Amount Raised: R540 000
Awareness: 45 000 people

#Raise4Rare

Rare Diseases South Africa



Total Raised through GivenGain:
R1 241 099
Fundraisers: 20
Campaigns: 27
Donors: 1021

givengain

Donate now!

IMPACT

Number of Projects: 11
Total Amount Raised: R738 715
Awareness: 45 000 people

R.A.R.E ACTIVists

Encouraging members of the public to ride /run / swim for a purpose, by using their mobility to benefit those without it, through FUNdraising and creating awareness.

Goals:

- Raised Funds for RDSA
- Create awareness and educate members of the public about Rare Diseases
- Brand Awareness
- Provide friends and family of rare disease patients, a platform to turn their feeling of helplessness into something positive that benefits our rare community.

IMPACT

- **Fundraising** totalling R4 691 000
- **Recruited** over 1500 Rare ACTIVists
- Raised **awareness** to over 210 000 South Africans

Impact Summary

Advocacy

- Rare Diseases Forum for private sector
- Improved collaboration
- Strengthened Advocacy
- Increased visibility
- Increased capacity
- Local & International participation
- United Voice
- Increased capacity
- Educated stakeholders

Education

- Improved patient centricity
- Educated all stakeholders
- Sharing of best practice
- Connected community
- Provided networking opportunities
- Improved knowledge base
- Improved referral pathways
- Increased patient support

Community

- Sharing of knowledge
- Improved patient resources
- Connected community
- Motivated patients
- Reduced stigma
- Allowed personal giving from the public
- Provided networking platform
- Raised awareness
- Develop Key messaging

Capacity

- Strengthened patient advocacy groups
- United voice
- Improved collaboration
- Prevent Burnout
- Improved sustainability
- Empowered patient advocates
- Improved knowledge
- Increased capacity across the healthcare sector

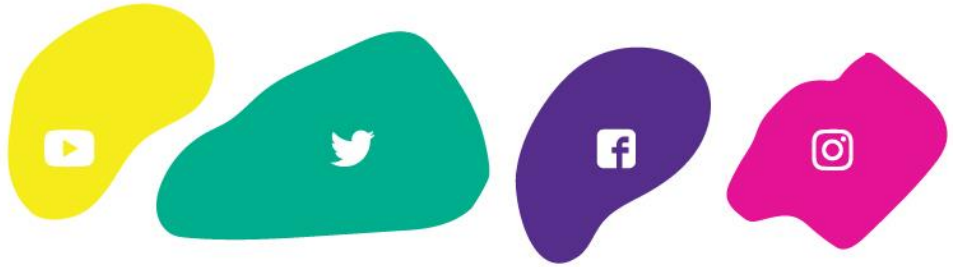
Awareness

- Raise awareness on pertinent challenges on a peer-to-peer scale
- Increase Awareness and visibility
- Encourage critical mass
- Improved understanding and knowledge base
- Increased patient – centricity
- Shared best practice
- Improved patient navigation

ACTIVists

- Increased awareness
- Increased visibility
- Increased Funding
- Improved Brand awareness
- PR and Media exposure
- Opportunity for personal giving
- Increased networking
- Community engagement
- Reduce stigma

Want to get involved?



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