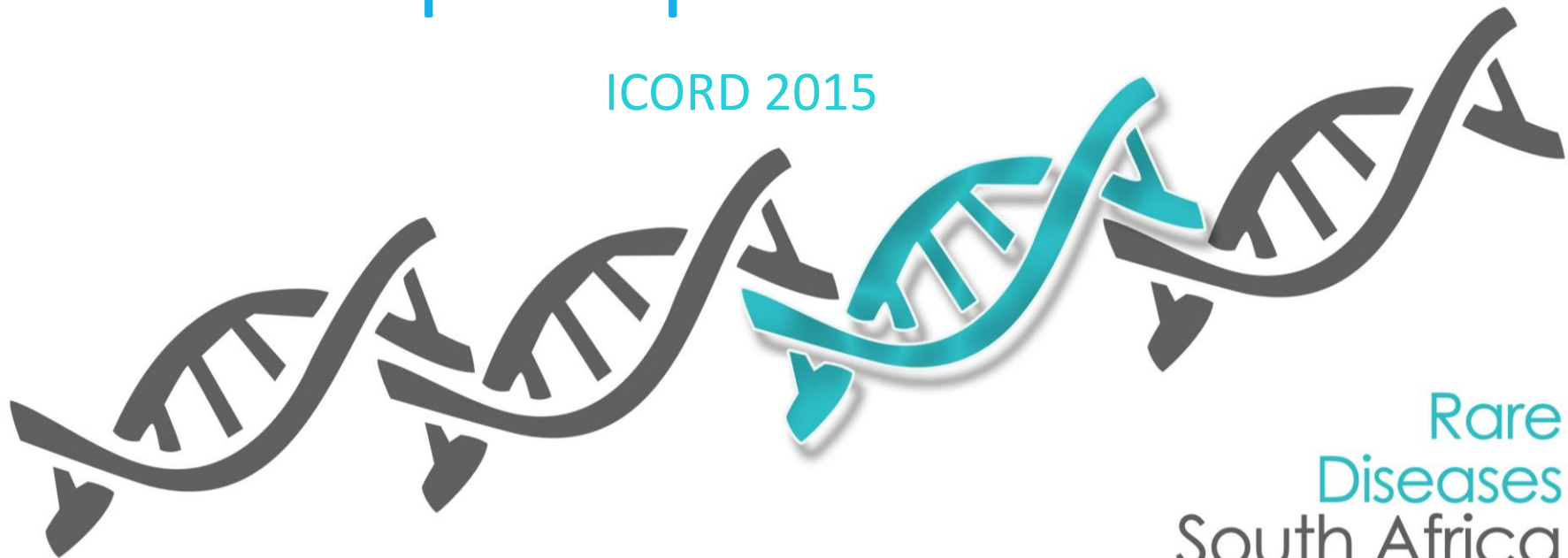


# Rare diseases from a parents perspective

ICORD 2015



Rare  
Diseases  
South Africa



## Conflicts of interest

As an organization, we are largely dependant on the donations from both public contributions and Corporate companies.

We do not allow any contributions to influence our aims and operational objectives.

All opinions in this presentation are my own.





## RDSSA – patient organization background

RDSA is a registered NPO which aims to ensure all patients with rare conditions in South Africa receive access to life-saving treatment and improved quality of life.

Founded by Kelly du Plessis, from personal need.

At that point, there was no organization in South Africa serving the needs of all patients with Rare Diseases, and hence its formation.

There are an estimated 3.7 million people with Rare Diseases in South Africa, with most being invisible in the healthcare system.



# Meet our family

Juan, diagnosed with Pompe in 2010, and then with a very rare additional glycogenosis PRKAG2 in 2013, really is a unique little boy

Now aged 6, Juan is a happy boy with a high quality of life. His sister, Jordyn, aged 4, is a healthy girl who adores her brother.





## Impact of rare diseases on family

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.

There are devastating repercussions affecting every sphere of life for Rare patients. The whole family is impacted.





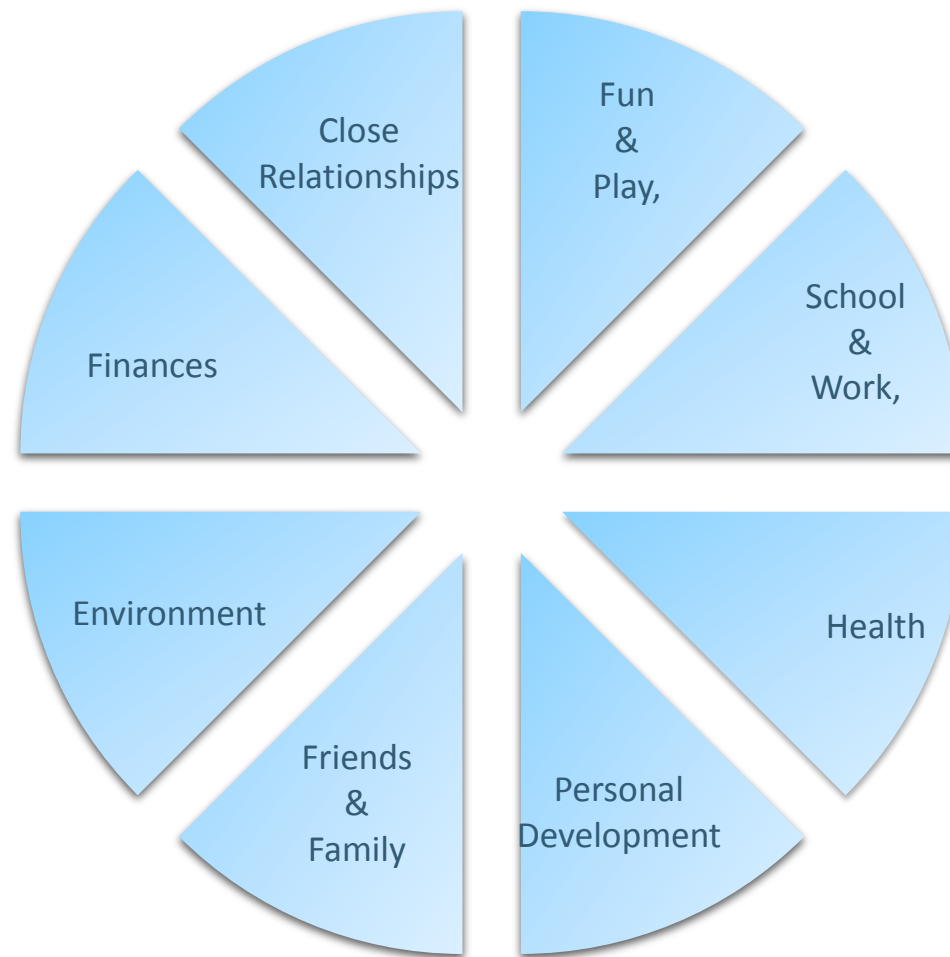
# Impact of rare diseases on family

The absence or confirmation of diagnosis with Rare Disease of a loved one leads to:

- Guilt/self-accusation
  - Divorce
- Limited Work opportunity
  - Social isolation
- Depression, Anxiety, Stress
  - Stigmatization
  - Financial burden



# Social Impact of rare diseases on family



# Everyone is different

*I found that families don't have feelings.... Individuals do. My feelings about this were different from my wife's, and those are different from my daughter's.*

*Everyone has their own, very individual experience. That has had an important impact on how our family has dealt with all of it. It's something that all families need to recognize when they are going through a shared experience like this.*

*Just because you feel or react one way, doesn't mean your wife or children are experiencing the same thing in the same way. It was quite a thing to realize.*

Hollaway 2007

# Everyone is different





# Death is a reality

- *Many rare conditions lead to premature death which has long term affects on those left behind.*
- *Even for conditions that are less severe, the uncertainty and isolation about the course of the diseases which has significant long-term impact.*

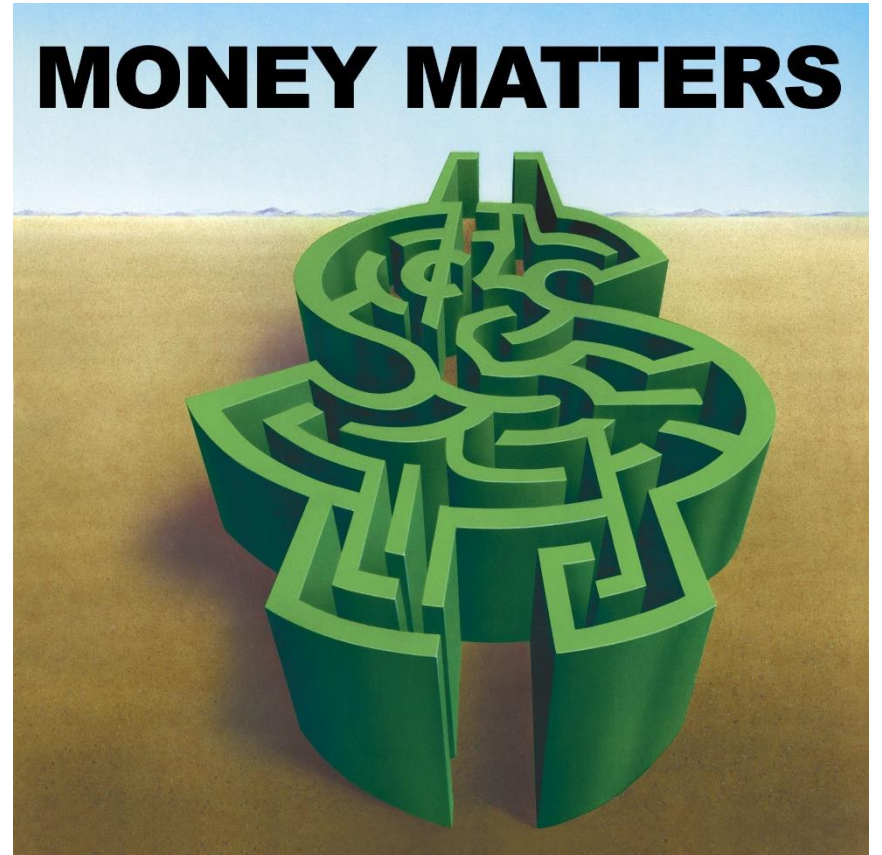
Death is not the  
greatest loss in  
life. The greatest  
loss is what dies  
inside us while  
we live.

-Norman  
Cousins



# Things to consider

- Associated costs with RDs include travel costs, medical aid insurance, time and money spent searching for diagnosis as well as treatment costs.
- RDs are long term diseases which can stretch over decades.
- Fulltime work is very difficult for both the patient and primary caregiver, generally reducing the total income of the household further straining the financial situation.
- Response to RDs are often shaped by socioeconomics, from difference in education and income status.



# Project Management

- Primary caregivers are often looked at as “Project Managers” – they are responsible for:
- making and attending appointments, treatments and hospitalizations,
- sourcing and administering of medication/supplements
- Ensuring patients is well fed, bathed, and cared for
- Communication between all stakeholders, doctors, insurance, support services,
- Payment and allocation of all medical bills and receipts
- Taking responsibility for the WHOLE picture



# We are just parents

- Somewhere in there, we are often still just parents!!
- Who need space to love, laugh, cry, hurt and just simply be a mom, or dad.
- This is still our child, and even though we have our “Project Management” hats on at most appointments, hearing bad news of regression, or uncertain futures, still hurts deeply.

We need the space to mourn the loss of our healthy children, and time to accept the rare child in their place.

# Hey, Mom!

You are allowed to be human.

You are allowed to be imperfect.

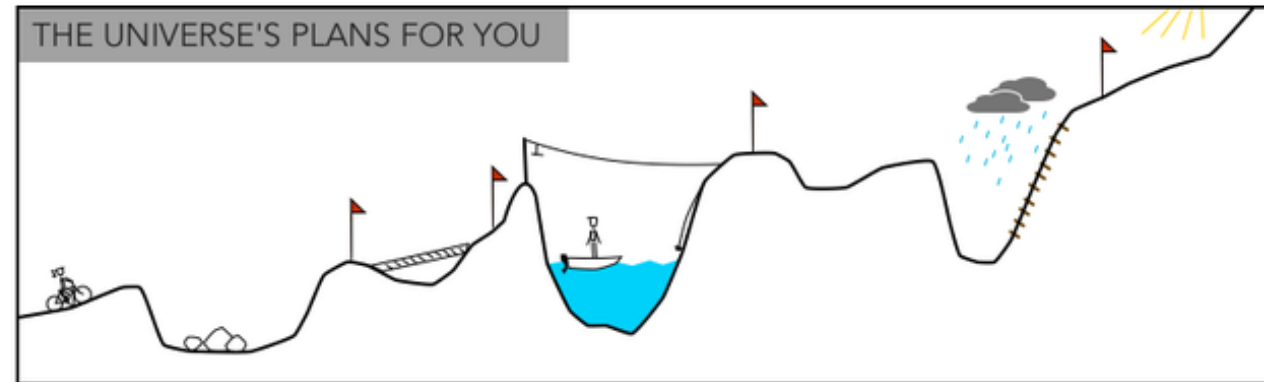
I am watching how you respond to real life situations and I am learning. Smile!

Love, Your Child



# Some days are worse...

- Some days can be overwhelming and traumatic.
- Establish the fundamental essentials that cannot be done without, and on these days only do those.
- The uncertainty of not knowing how long the “race” is and when to expect hills is a major contribution to fatigue. Accept that certain things cannot be changed.
- Don't plan to achieve, plan to cope.
- Coping means surviving.



DOGHOUSE DIARIES



Rare  
Diseases  
South Africa



# Reminder to Caregivers

You can't pour from an empty cup.  
Take care of yourself first.





# Siblings

- Siblings, siblings, siblings....
- Often the focus is solely on the sick child leaving the siblings with the same feelings as the patient:
  - -isolated
  - Forgotten
  - Hurting
  - Ignored.
- However, unlike with the patient, these feelings are often not recognized or understood by others in the family.

The lack of understanding and recognition regarding how they feel does not negate their feelings.

Siblings often feel guilty for feeling this way. Siblings should be allowed to swing back and forth between positive and negative emotions as parents do.

"Is it fair?"

being a sibling of a child with special needs



# Siblings

- Siblings are also impacted by:
  - Lack of available time from parents and family
  - Lack of available funds for their wants and desires
  - Needing to change their schedule to suit the sick sibling
  - Not being able to plan their own days
  - An immense sense of responsibility over their sick sibling
- They need to be supported and recognized as being a part of the solution, and empowered from a young age to contribute to the family dynamics, as opposed to just being included as a member of the family



# Conclusion

- Problems affecting our families are unique  
But so are our children, and with the unique  
problems comes unique love.

*Would we change it? Never!*



THANK YOU



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