How to Care for the Rare

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MOTHER OF A CHILD WHO IS RARE

Objectives

Provide Insight into the Rare Diagnosis

Tailoring of Care for the Rare

How to find joy amidst the challenge

Disclosures

I have no disclosures

My Story

Family of 4

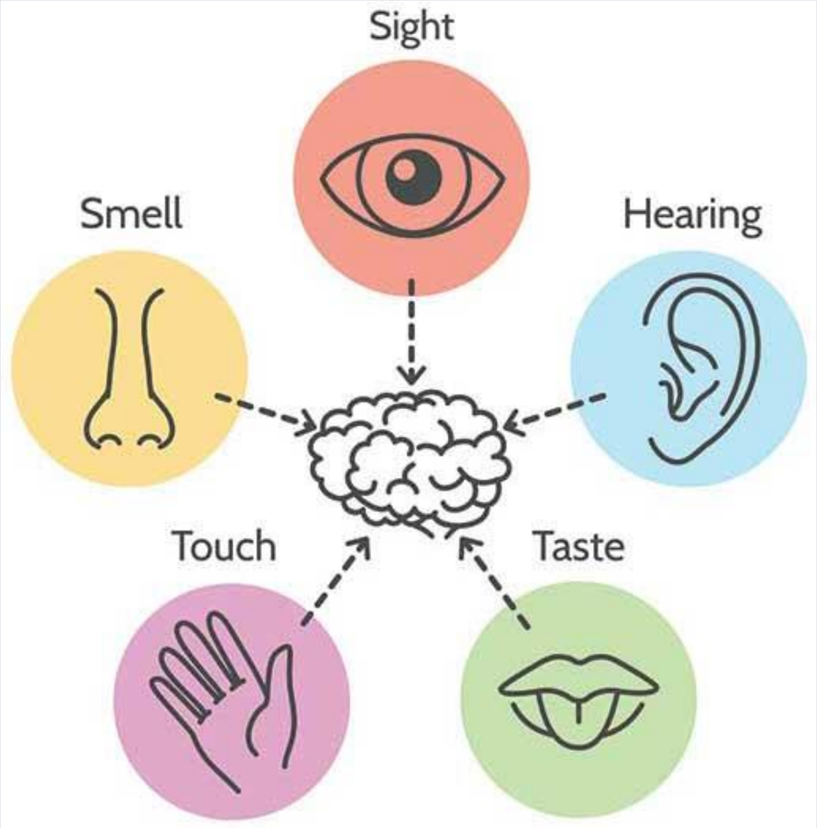
Nora 3 ½

Aubrey 18 months

Longing for one more

Big Life Choice

Premonition?



About 20 weeks pregnant

Random windshield time solo with husband

Which sense would you choose to lose?

Something felt wrong

Meet Mac

- Spontaneous bilateral Pneumothoraxes
- Optic Nerve Hypoplasia, bilaterally
- Polymicrogyria
- Frontal Lobe Migrational abnormalities

First Year

Lonely

Guilt

Anger

Sadness

Fear

Inadequate

Understanding Your New Life

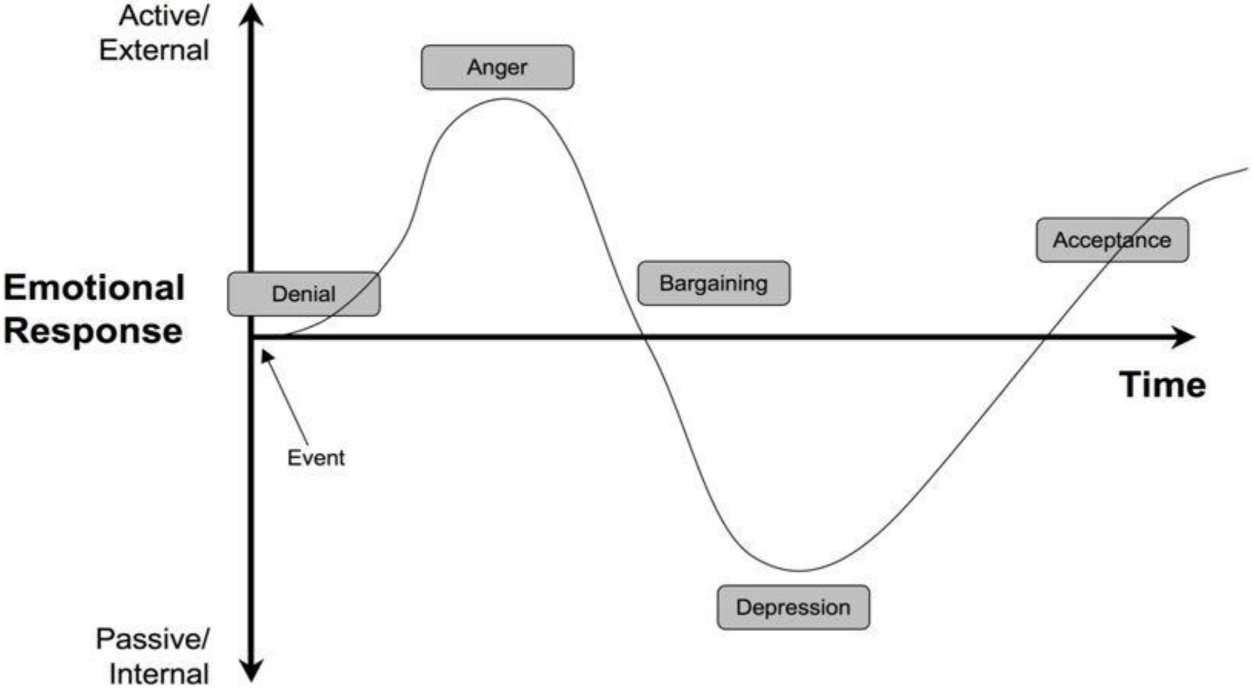
The loss of normal

So many appointments

No one is the expert

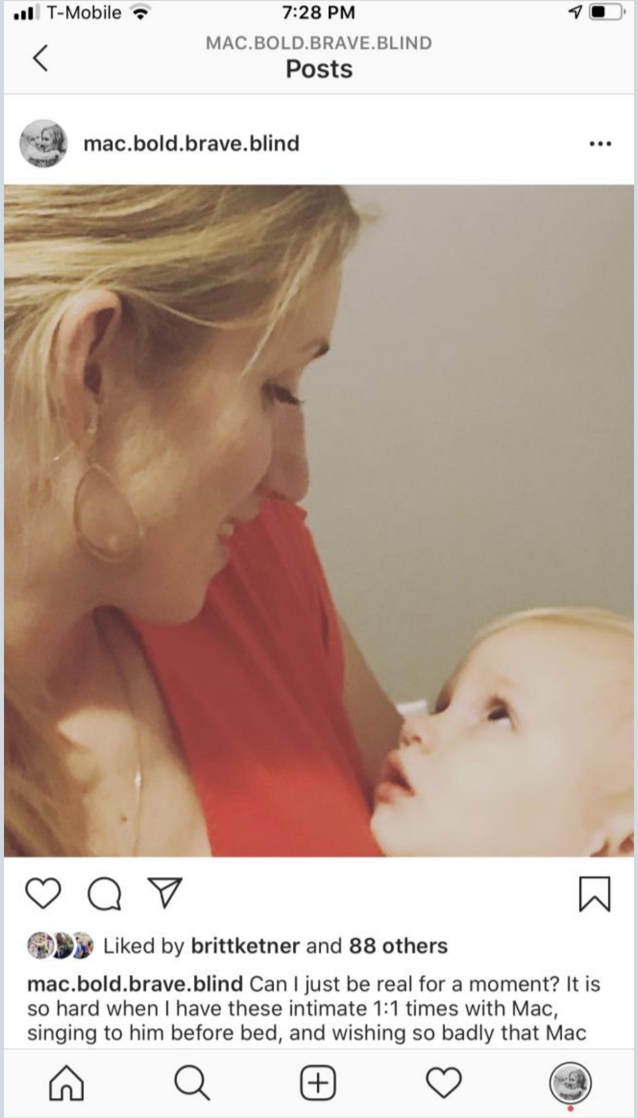
Pretending it’s all okay

5 Stages of Grief Cycle



It’s okay not to be okay

- We don’t always understand what will break us down
- Remember each person is on a unique journey
- Grieving the loss of normal is healthy
- Don’t say “it could be worse”



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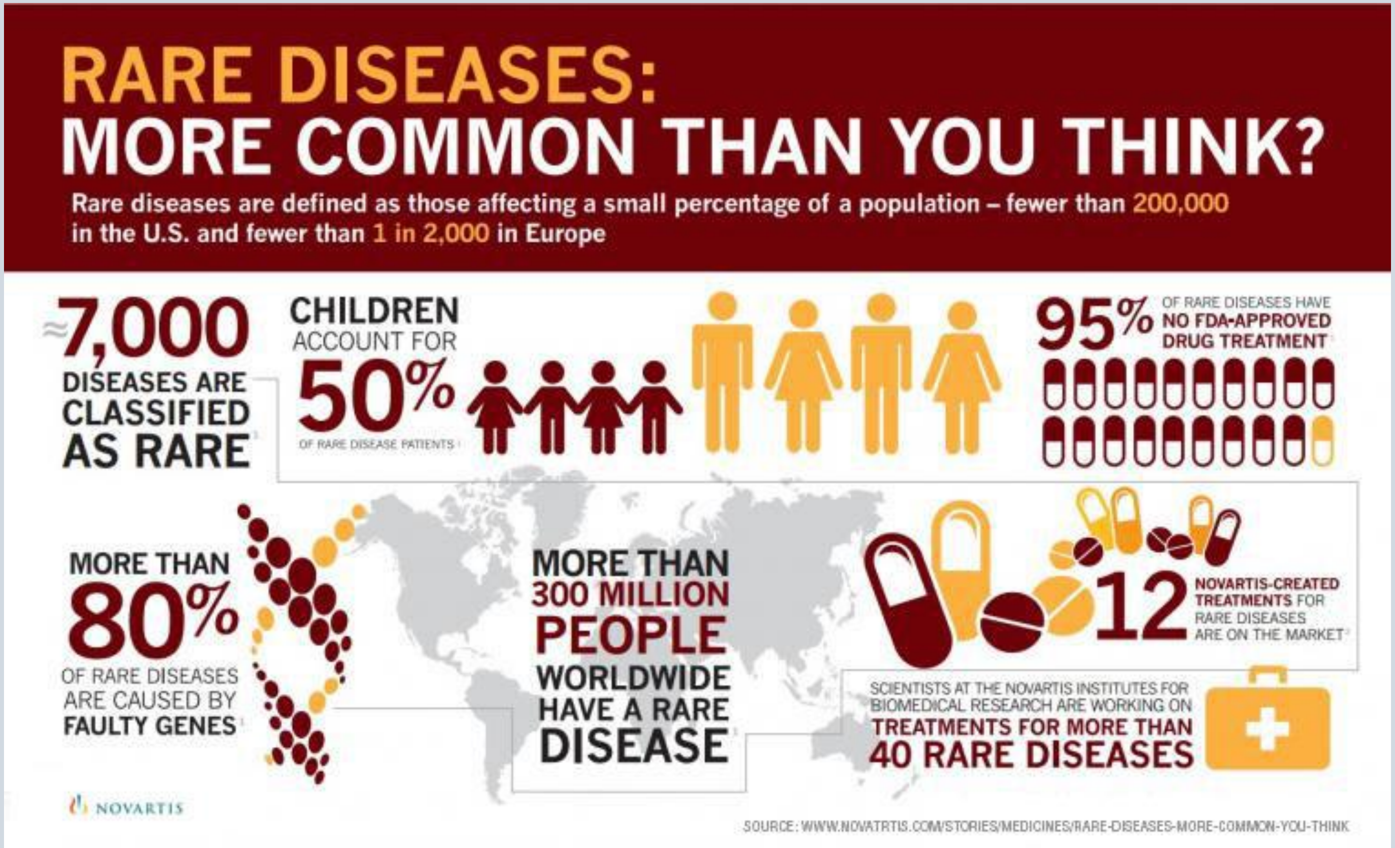
mac.bold.brave.blind Can I just be real for a moment? It is so hard when I have these intimate 1:1 times with Mac, singing to him before bed, and wishing so badly that Mac

Helpful Hints

- Say hello to the child
- Ask the parent/caregiver how they are doing
- Acknowledge their journey
- Don’t focus on the disability
- Don’t make assumptions
- Avoid showing pity
- Don’t say “it could be worse”

Rare Disease Facts



RARE DISEASES:
MORE COMMON THAN YOU THINK?

Rare diseases are defined as those affecting a small percentage of a population – fewer than 200,000 in the U.S. and fewer than 1 in 2,000 in Europe

≈7,000 DISEASES ARE CLASSIFIED AS RARE

CHILDREN ACCOUNT FOR 50% OF RARE DISEASE PATIENTS

95% OF RARE DISEASES HAVE NO FDA-APPROVED DRUG TREATMENT

MORE THAN 80% OF RARE DISEASES ARE CAUSED BY FAULTY GENES

MORE THAN 300 MILLION PEOPLE WORLDWIDE HAVE A RARE DISEASE

12 NOVARTIS-CREATED TREATMENTS FOR RARE DISEASES ARE ON THE MARKET

SCIENTISTS AT THE NOVARTIS INSTITUTES FOR BIOMEDICAL RESEARCH ARE WORKING ON TREATMENTS FOR MORE THAN 40 RARE DISEASES

NOVARTIS

SOURCE: WWW.NOVATRTIS.COM/STORIES/MEDICINES/RARE-DISEASES-MORE-COMMON-YOU-THINK

Did you know…

- 80% of parents of children with special needs end up divorced
- The financial burden on families can be so overwhelming and debilitating
- For 95% rare disorders there is not a current FDA approved treatment – and for some, the therapy is not covered by insurance.
- Approximately 50 percent of patients affected by rare diseases are children and 30 percent of these children will not live to see their fifth birthday.

How to Care for the Rare

Realize the parents are a member of the healthcare team

It’s okay not to have the answers

See the patient as a person

Take a moment to be authentic

KNOW YOURSELF.

BE AUTHENTIC.

HAVE COURAGE.

Caring for the Rare: Our Journey Changes

No light to Guide Us

New diagnoses:
  - Congenitally Absent ACL’s
  - Aortic Root Dilation

Current Learnings:
  - Never know the path
  - Waves keep coming
  - Let others help

From Heartbreak to Joy

Life Takeaways

Slow down

Don’t make assumptions

Find the joy

Enjoy the little things

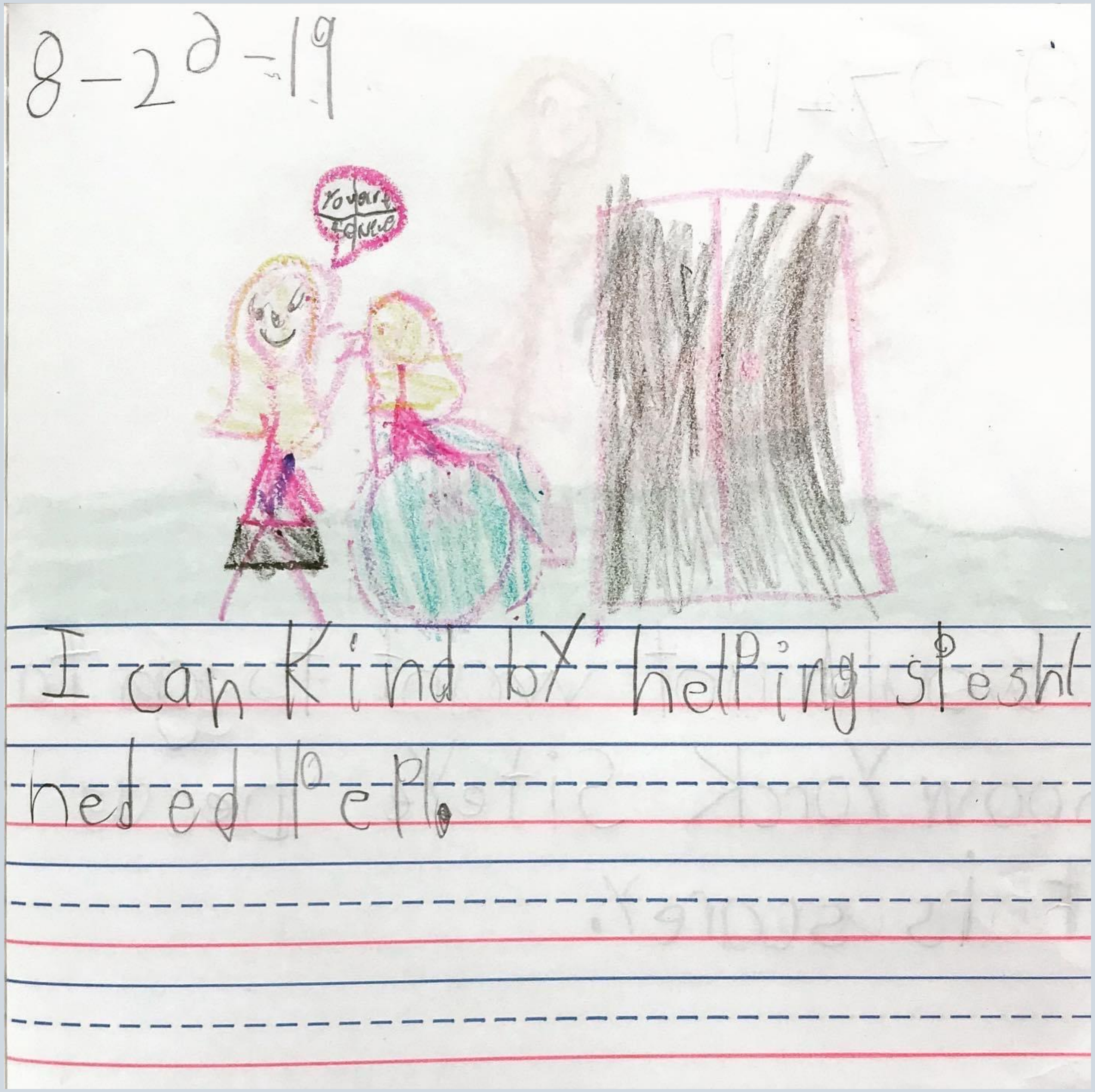
What really matters?

Everything has beauty,
but not everyone sees it.

Confucius

Perseverance: persistence in doing something despite difficulty or delay in achieving success

Takeaways



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Everyone’s journey is unique

Celebrate the victories

Allow yourself to grieve your loss of normal

Support others in their journey

Thank you! Any Questions?

References

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