

A quick read before we start...

Welcome to Holland

by Emily Perl Kingsley

I am often asked to describe the experience of raising a child with a disability – to try to help people who have not shared that unique experience to understand it, to imagine how it would feel. It's like this.....

When you're going to have a baby, it's like planning a fabulous vacation trip – to Italy. You buy a bunch of guide books and make your wonderful plans. The Coliseum. The Michelangelo David. The gondolas in Venice. You may learn some handy phrases in Italian. It's all very exciting.

After months of eager anticipation, the day finally arrives. You pack your bags and off you go. Several hours later, the plane lands.

The stewardess comes in and says, "Welcome to Holland." "Holland!?" you say. "What do you mean Holland?? I signed up for Italy! I'm supposed to be in Italy. All my life I've dreamed of going to Italy.

"But there's been a change in the flight plan. They've landed in Holland and there you must stay.

The important thing is that they haven't taken you to a horrible, disgusting, filthy place, full of pestilence, famine and disease. It's just a different place.

So you must go out and buy new guide books. And you must learn a whole new language. And you will meet a whole new group of people you would never have met. It's just a different place. It's slower-paced than Italy, less flashy than Italy.

But after you've been there for a while and you catch your breath, you look around.... and you begin to notice that Holland has windmills....and Holland has tulips. Holland even has Rembrandts.

But everyone you know is busy coming and going from Italy... and they're all bragging about what a wonderful time they had there. And for the rest of your life, you will say

"Yes, that's where I was supposed to go. That's what I had planned."

And the pain of that will never, ever, ever, ever go away... because the loss of that dream is a very very significant loss.

But... if you spend your life mourning the fact that you didn't get to Italy, you may never be free to enjoy the very special, the very lovely things ... about Holland.



My Pompe Journey

Presented by: Jennie Giusto
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Facebook Group: Pompe Strong

A little about me...

I have been teaching Kindergarten for 15 years. I am married with two children, Sophia and Angelina. Sophia is five years old and Angelina is 15 months old. We live in Long Island, NY.



Sophia, Our Pompe Warrior



Sophia was born in 2015 and was diagnosed with Late Onset Pompe Disease on the NY Newborn Screening.

Pompe Disease had only been added to this screening a year prior to her being born. Also, at that time, Pompe was only found on two state screening panels; NY and Missouri.



The Call...

- Results from the newborn screening came back positive for Pompe Disease.
- Could be a false positive, don't panic.
- See a geneticist for a more definitive answer.
- Pediatrician was unsure what Pompe Disease was; couldn't give many answers.



The Aftershock: Stages of Emotions

01

“This can’t be happening.”

Shock and denial. This is a state of disbelief and numbed feelings

02

“Why Me?”

Anger and resentment.

03

“This is our fault.”

A lot of pain and guilt.

04

“How can I fix this?”

Helpless and hopeless. Uncertainty from doctors; unanswered questions.

05

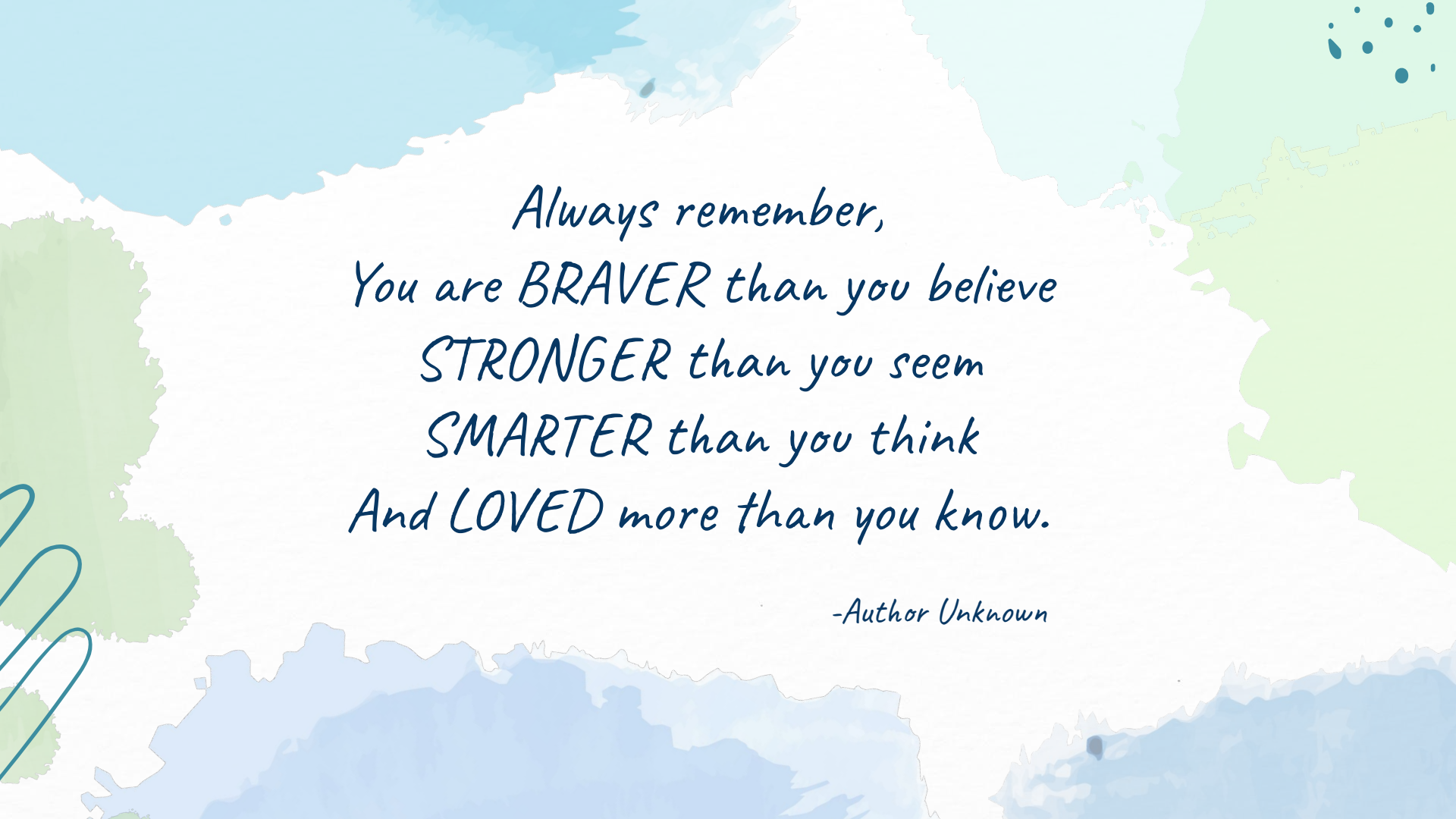
“I’m Lost”

Depressed and crying all the time. Difficulty enjoying being a mom. Unsure what to do next.

06

“I can do this!”

Accepting the diagnosis and moving forward. Finding hope and happiness in the little things.



Always remember,
You are **BRAVER** than you believe
STRONGER than you seem
SMARTER than you think
And **LOVED** more than you know.

-Author Unknown

Facebook Groups

Did You Know?

- Pompe Strong
- Pompe Alliance
- Moms of Pompe Kids
- Hope Travels - Pompe Awareness
- Pompe Caregivers Forum
- Cure Pompe Disease
- Pompe Friends

Our Pompe “Angel” Dr. Kishnani

Chief of Medical Genetics at Duke University
Durham, North Carolina



**November
2015**

Diagnosis & Duke

Met with Dr. Kishnani a few months after being diagnosed to get final guidance on how to proceed.

**August
2016**

First Port Placement

We decided to go with a port due to Sophia being 9 months old at the time.

**September
2016**

ERT

(Enzyme Replacement Treatment)

Begins

A very difficult decision to make but we have no regrets. Sophia gets bi-weekly infusions.

**Present
Day**

Home Infusions

After being monitored closely at the hospital for almost a year, we were able to transition to home infusions.

Fundraising & Raising Awareness

The Pompe Strong Team has raised over \$41,000 for Dr. Kishnani's Pompe Research!



#representrare

#pompestrong





**Just a
Typical
Kid!**



Things I've Learned...

- Take one day at a time! If you waste time worrying about what will come, you might miss out on what's important now.
- Enjoy the little things! The little smiles, the hugs and kisses...
- You are your child's biggest advocate! No one is going to care as much about your child as you are.
- Don't take no for an answer! Listen to your gut, follow your heart and do what is right for you and your family. Whatever decision you make is the right decision.
- You are stronger than you know! You can do it, and if you can't do it now, you will do it in time. You'll have good and bad days, but that's ok! You've got this!

*Welcome to Holland
There's no place like home...*



THANK YOU!