SPiCE

SPECIAL PEOPLE IN CATHOLIC EDUCATION

RARE DISEASES

Awareness • Support • Treatment • Research

February 2025

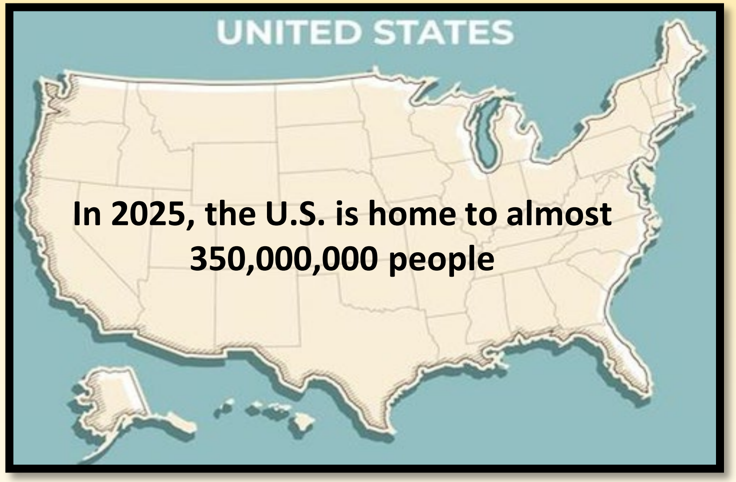
How is **RARE DISEASE** defined?

What does RARE mean? Not often found or seen; not common, unusual

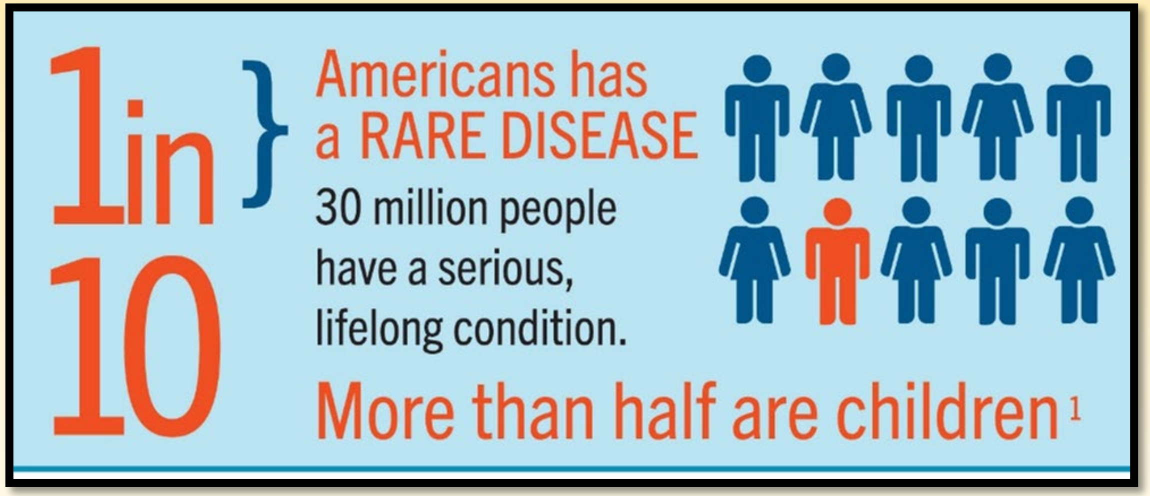
What is a DISEASE? A condition that affects the normal functioning of the body and causes illness

RARE DISEASE: A health condition that impacts a small percentage of the people

Any condition affecting less than 200,000 people in the United States.



In 2025, the U.S. is home to almost 350,000,000 people



Americans has a RARE DISEASE

30 million people have a serious, lifelong condition.

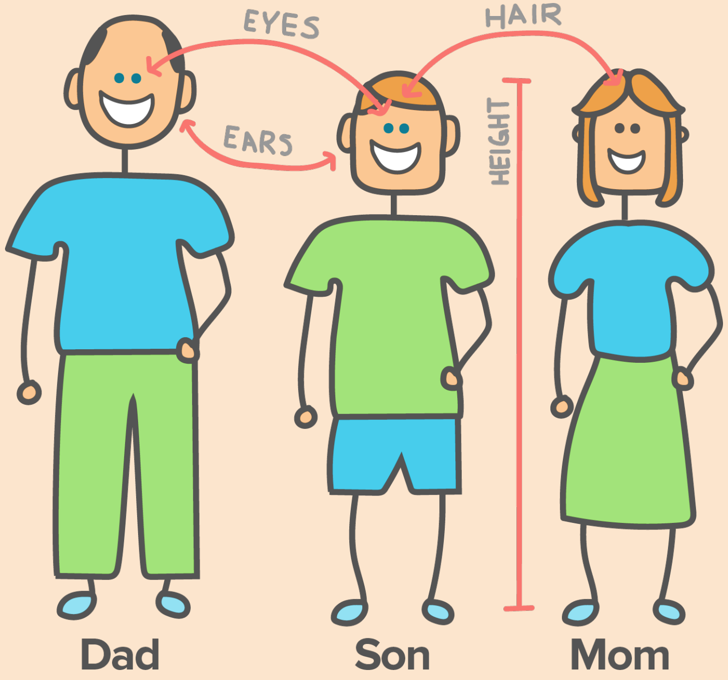
More than half are children[1]

Who is affected by Rare Diseases?

According to the National Institutes of Health (NIH): <u>there are over 7,000 Rare Diseases</u>.

The majority of rare diseases are *genetic*

Genes are passed down from parent to child, a special set of instructions that tell your body how to grow and work



Some rare diseases appear as a baby or young child, others do not appear until later in life.

What are some of the problems people with RARE DISEASE experience?

- Difficulty in gaining an accurate diagnosis, it can even take years!
- Little or no research being done on the disease
- Difficulty finding a doctor who knows how to treat the particular disease
- Paying for treatments that are generally more expensive than those for common diseases
- Feelings of loneliness
- Of the 7,000 known rare diseases, approximately 95% have no treatment

A Patient's Story: Paul's Story

This is Paul.

When Paul was just a toddler, he was diagnosed with Duchenne Muscular Dystrophy.

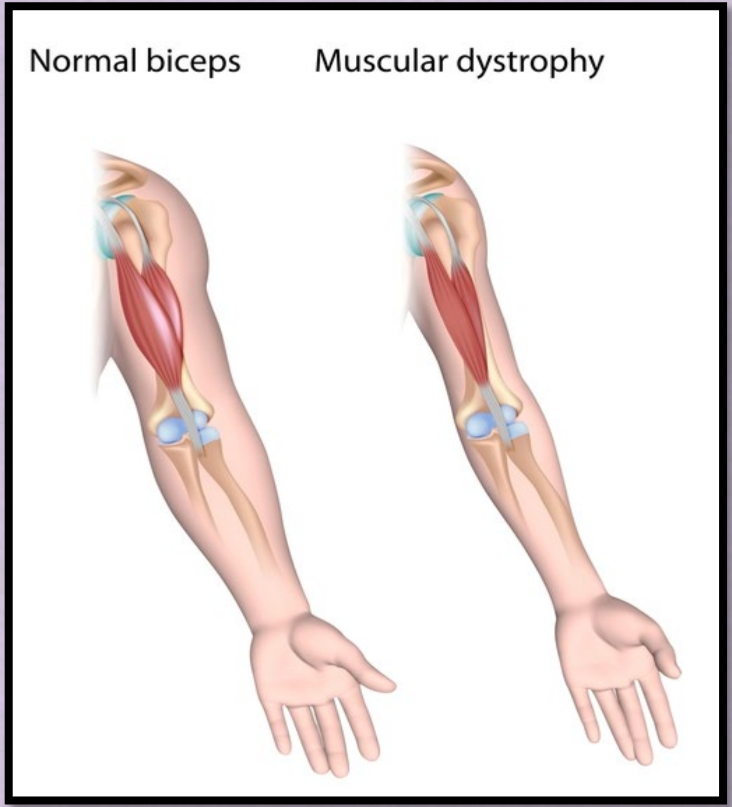
Duchenne Muscular Dystrophy is a rare genetic disease with no cure.

This disease affects people's muscles from a young age.

The muscles, including the heart muscle, get weaker and break down over time.

This makes it difficult for children to jump, run, and walk.

A Patient's Story: Paul's Story, continued



Paul feels lucky to receive the medical help and therapies he's received.

Paul is now 12 years old.

He enjoys model cars and video games with his brother.

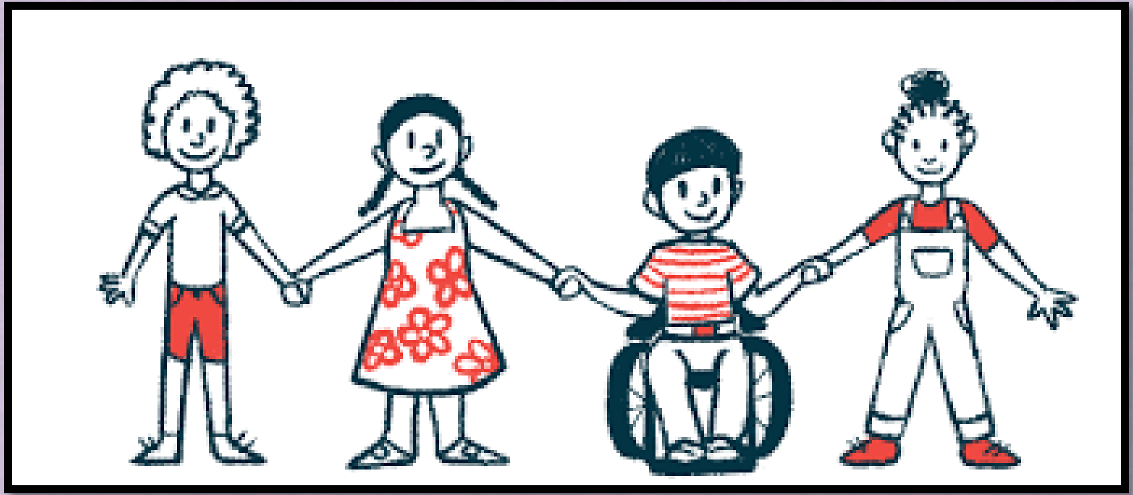
Unfortunately though, raising his arms has become much more difficult.

He is unable to stand or maintain his balance.

A Patient's Story: Paul's Story, continued

Paul needs support for all daily activities, including getting in and out of bed. His muscles have broken down so much, that he must now use a wheelchair for the rest of his life.

Paul's parents help spread awareness about the difficulties patients and their families experience from the moment their child is diagnosed with a rare disease.

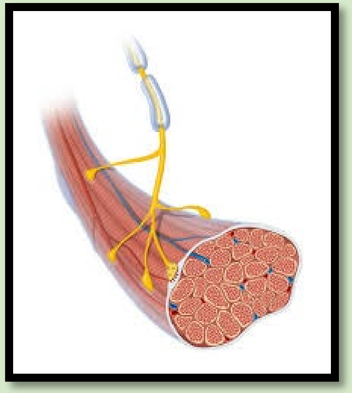


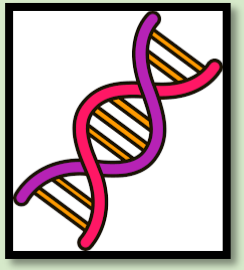
A *Rare* Name for A *Rare* Disease

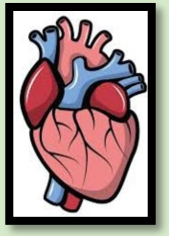
Many Rare Diseases are named for the physicians who first discovered them.

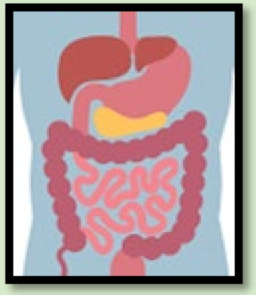
A few are named for patients, the hospitals where they were first identified, even the location of the body they affect. These locations include:

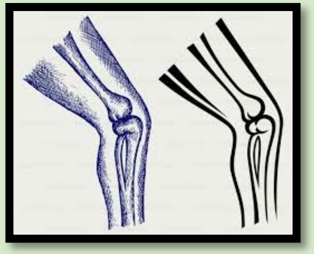
- Neurological (brain) and neuromuscular (nerves/muscles)
- Metabolic (digestive/stomach)
- Chromosomal (DNA/genes)
- Skin and bone
- Heart, blood, lungs, kidneys, and other body organs and systems

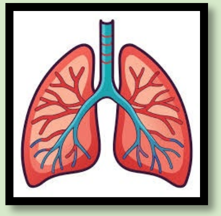












A Parent’s view

Each day I wake up, I pray that God has not taken (my son) away from us in the night.

When I lay eyes on him, and see his smiling face, it lights up my world.

Some days (my son) wakes up cheery and bouncing.

Other times he wakes up groggy because it's been a hard night.

Changing his diaper from over night is always different, sometimes its so full that it has leaked, sometimes its (yucky) and a red bum, and sometimes its just a quick change.

Getting (my son) changed and clothes on for the day, which is a challenge too.

Because clothes that fit my 9 year old is not made for diapers, it's not always accessible, and sometimes they don't work with his braces, walker, or shoes.

A Parent’s view, continued

Then (my son) is lifted into his chairlift, so he can get down the stairs safely.

This is new, because we have always carried him down the stairs.

But at 50 lbs and nearly 4 1/2 ft tall, it was a huge hazard.

When we get downstairs, (my son) sits in a special chair for meals and snacks.

I put on some cartoons for (my son) while I get his meds ready.

Each syringe, each bottle is specially marked.

I know each bottle and each dose…

Make sure that you are precise on the measurements…

All the meds total $35,000 each month…

A Parent’s view, continued

The first hour is an exhausting start to the day.

But it's an important time to get everything on our list checked off, to keep (my son) safe.

We fight to find doctors who will listen.

We fight insurance companies when they deny tests, treatments, and therapies.

We fight the education system that is not prepared for our rare child.

We fight each day because at the end of the day that fight is life changing for our loved one.

RARE DISEASE DAY

An awareness event that takes place every year

on the last day of February:

February 28

(or February 29 during Leap Years)

The rarest of calendar dates to emphasize the nature of these diseases

Dr. Meghan Martin, a pediatric emergency medicine physician who shares medical ideas for families via social media platforms.

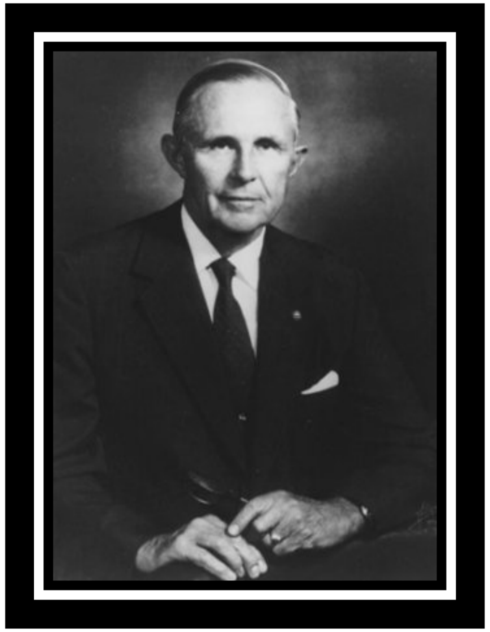
Click to view the video she shared on Rare Disease Day last year.

Did you know?!

- In the United States, zebras are more rare than horses.
- Every zebra has a unique stripe pattern, like fingerprints, no two patterns are alike.

- *The term* ***"ZEBRA"*** *is often used to describe a rare or unexpected diagnosis.*

… But how did "Zebra" become a synonym for Rare?



Horse or Zebra? Hooves Matter

Dr. Theodore Woodward, a professor in the 1940s, told his students:

"When you hear hoofbeats behind you, don't expect to see a zebra.“

Did you catch this similar phrase, referenced in Dr. Megan Martin’s video, on one of the previous slides?

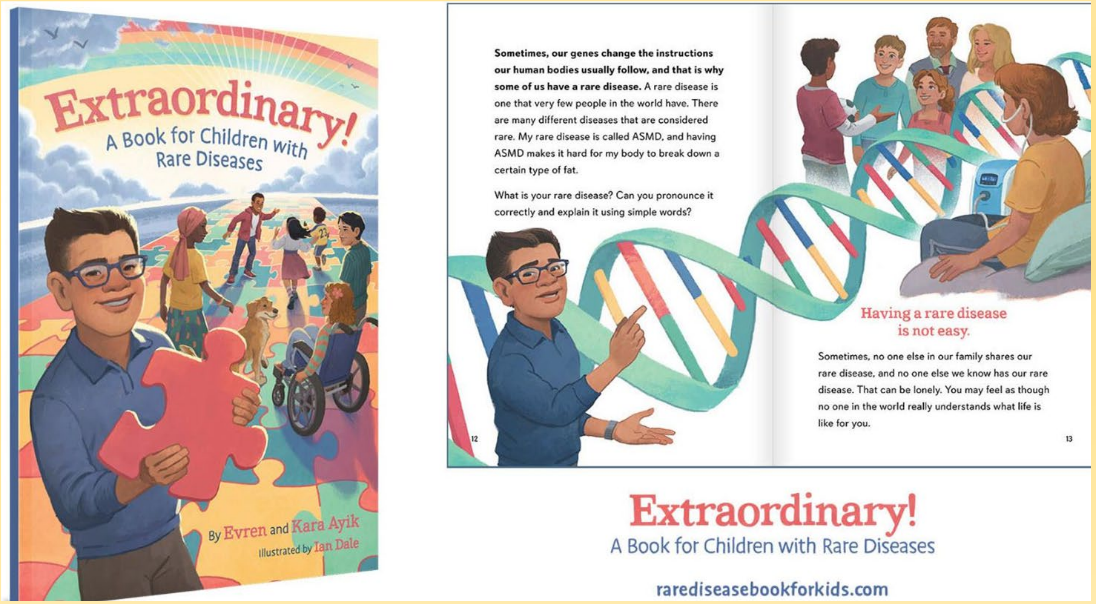
Dr. Woodward used this metaphor to teach the diagnosis of diseases.

Horses are much more common animals, so assume it is a horse behind you, rather than a zebra.

In medical terms: think of the common diseases before the uncommon diseases.

With this information in mind, it should be no surprise that the *ZEBRA* is the mascot for rare diseases!

Book Recommendations



Extraordinary!
A Book for Children with Rare Diseases
By Evren and Kara Ayik
Illustrated by Ian Dale

Sometimes, our genes change the instructions our human bodies usually follow, and that is why some of us have a rare disease. A rare disease is one that very few people in the world have. There are many different diseases that are considered rare. My rare disease is called ASMD, and having ASMD makes it hard for my body to break down a certain type of fat.

What is your rare disease? Can you pronounce it correctly and explain it using simple words?

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Having a rare disease is not easy.

Sometimes, no one else in our family shares our rare disease, and no one else we know has our rare disease. That can be lonely. You may feel as though no one in the world really understands what life is like for you.

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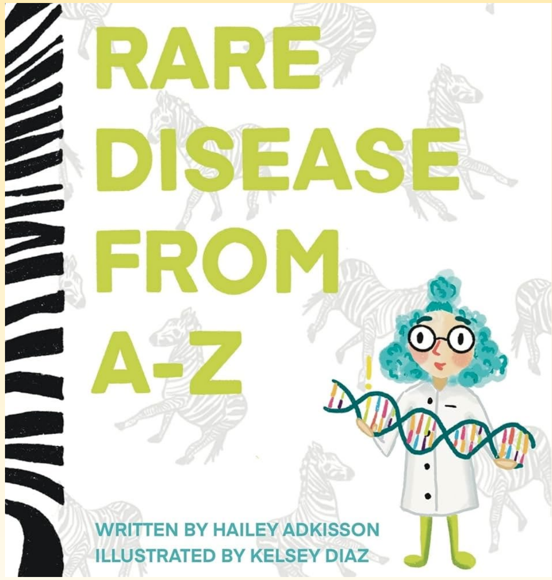
Extraordinary!
A Book for Children with Rare Diseases
rarediseasebookforkids.com

Written collaboratively by mother and son,

Extraordinary! A Book for Children with Rare Diseases

opens up a child-friendly discussion about identity, inclusion, and self-concept in light of the challenges and silver linings of living with a rare disease.



RARE
DISEASE
FROM
A-Z

WRITTEN BY HAILEY ADKISSON
ILLUSTRATED BY KELSEY DIAZ

B Baraitser Winter Syndrome

The symbol for BWS is a turtle, for a slow and steady path, and progress at their own pace.

Baraitser-Winter Syndrome (BWS) is a rare genetic change on the ACTB or ACTG1 genes that affects the growth of many parts of the body, especially the face and brain. Intellectual disability is very common. Many people with BWS have similar faces including widely spaced eyes, droopy eyelids, a wide nose, and big, wonderful smiles.

Most individuals with BWS have some level of hearing loss.

BWS impacts how the brain forms and grows and can cause agenesis of the corpus callosum.

Seizures are very common.

BWS can cause problems with the kidneys and the heart.

Short stature (height) is also common.

Rare Disease From A-Z

Every page gives a description of the disease as well as how rare it is… While not every disease will have images of children on them, the ones that do are based off of real kids with the same diagnosis!

It's very important to us that we teach kids (and adults) real terminology, while explaining it in a simple way.

SHOW YOUR STRIPES!

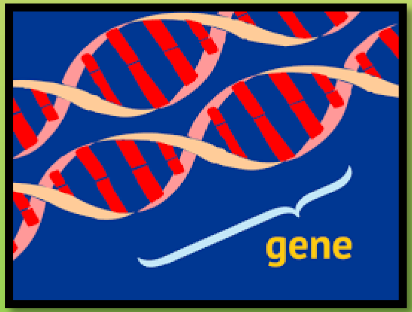
St. Brigid students and faculty will be

Celebrating Rare Disease Day on **<u>Friday, February 21st</u>**

Students are encouraged to ***dress down,*** wearing

Jeans

on this day, to raise awareness of the daily challenges faced by those living with **genetic rare diseases**.



Let's see your support with jeans for genes!

The SPiCE Committee of SBK appreciates your support!

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