SCIENCE FAIR LOGBOOK 2024-2025 STUDY PROJECT. PROGERIA: WHAT IS IT? TREATMENTS, SYMPTOMS, AND POSSIBLE CURES.

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Important dates:

- October 22nd: Log into CYSF platform
- Nov 8 2024: decide topic
- Dec 3rd 2024: check in before winter break
- Jan 6th, 2025: trifolds are available
- Jan 20th, 2025: finish and bring in trifolds

What is progeria?

Progeria, also known as Hutchinson Gilford Progeria Syndrome (HGPS) is a rare genetic disorder that is caused by a genetic mutation in the Lamin A gene. This gene makes lamins which are genes that line the inside of the nuclear membrane so that there is a platform to build chromatin and protein. But, when you have progeria, instead of making lamins, this gene makes progerins which are genes that make your skin shrivel up and make you look like and old person. Progerins also cause DNA damage by disturbing replication of DNA. This disease usually affects young children and the signs start to show at around 2-3 years old. Children with progeria age about 8-10 times faster which makes them not live past 14 years usually.Everyone has progerins in their bodies, but people with progeria make a lot more progerin causing them to age faster.

Scource: Link

More about HGPS

The stress caused by progeria is dected by special proteins like c-gas and STING that activate a type of immune response that leads to declining cells. But, this response is activated without the release of mollecules like interferons that usually trigger your immune system. Most people did not know about HGPS until Adalia Rose, a youtuber with progeria started posting on the internet. People then became more aware of this disease after Adalia passed away at the age of 15. Heart problems or strokes are eventually the cause of death in most children with progeria. It is quite common for kids with progeria to get heart attacks and strokes, which can be dangerous and be fatal.

Scource: Link

Treatment

Progeria can be treated with calcitorl, a type of vitamin D that rescues more cells from the glitches caused by progerin, making you gain more normal cells than progeria cells over a long period of time. We still need to look deeper into this drug. Who knows? Maybe we could extract a cure for progeria from calcitriol one day!

Some medication for progeria includes:

- Lonafarnib (Zokinvy): this medication helps to slow down the buildup of progerin
- Low dose asprin: daily dose might help prevent heart attack and stroke
- Physiotherapy & other treatments: This helps stiffen joints, you can get hearing aids for hearing loss, vision therapy for eyesight loss, and many more treatments.

Scource: Link Video

Symptoms of Progeria

Signs of progeria can start showing shortly after the child is born and when you go to your pediatrician for checkups, they will tell you if your child has any symptoms of progeria.

Some symptoms of progeria include:

- Large head
- Protruding eyes
- Skin that looks old and wrinkly
- Pinched nose with point at the end
- Ears that stick out
- Visible veins on the scalp, forehead, or between the eyes
- A high pitched voice
- Skinny (loss of body fat and muscle)
- Tight joints (usually cause pain)
- Hair loss (including eyelashes and eyebrows)

Scource: Link

The oldest person with progeria

Linda Wedekind, a 73 year old woman with progeria is the longest living person with progeria. She was never diagnosed with progeria as it was so mild that you cant tell. Her children, Chad and Tiffany Wedekind were both diagnosed with progeria and Chad unfortunately passed away. Tiffany, however is still alive and is currently 45 years old as she was unusually diagnosed with progeria in her late 20's. (more about them later)

Scource: Video Video

Vocab

Lamin

- Lines inside of nuclear membrane, providing a platform for protein and chromatin binding.

Chromatin

- Gene that protects your DNA and controls your cell cycle

C-Gas

- Detects cytosolic DNA
- Cytosolic DNA
- Water based fluid with cellular structures like protein inside it. It indicates cellular damage or infection, triggering your immune system.

Scource: Wikipedia

Vocab continued

STING- stimulation of interferon genes

Interferon- genes that usually detect viruses and other bad things for your body and trigger your immune system.

Scource: Wikipedia

Some people with progeria

ADALIA ROSE: FAMOUS YOUTUBER WITH PROGERIA. DIED AT THE AGE OF 15



TIFFANY AND LINDA WEDEKIND: OLDEST PEOPLE WITH PROGERIA AT 73 AND 44 YEARS OLD YEARS WITH PROGERIA



Case Studies

Adalia Rose- A shining star



ADALIA ROSE WILLIAMS WAS A SUPER INSPIRING GIRL WHO BECAME VERY FAMOUS ONLINE FOR HAVING A RARE CONDITION CALLED HUTCHINSON-GILFORD PROGERIA SYNDROME, WHICH MADE HER BODY AGE 8-10 TIMES FASTER THAN NORMAL. BUT INSTEAD OF LETTING THAT HOLD HER BACK, ADALIA LIVED AN AWESOME LIFE WITH LOTS OF ENERGY.

ADALIA WAS BORN ON DECEMBER 10, 2006, IN TEXAS AND SHE WAS DIAGNOSED WITH PROGERIA AS A 3-MONTH OLD BABY.EVEN THOUH SOME PEOPLE WOULD STARE AND BE MEAN, SHE BRAVED THROUGH ALL OF THE HARDSHIPS.

- LINK: ADALIA ROSE
- LINK: ADALIA ROSE PROGERIA

ADALIA ROSE- A SHINING STAR

ADALIA AND HER MOM STARTED POSTING VIDEOS ON YOUTUBE WHEN SHE WAS LITTLE, AND PEOPLE INSTANTLY LOVED HER. SHE MADE MAKEUP TUTORIALS, SHOWED OFF HER BIRTHDAY PARTIES, AND EVEN DID HILARIOUS DANCE VIDEOS. HER CATCHPHRASES LIKE "SLAY THE DAY AWAY!" MADE HER EVEN MORE POPULAR AND FUN TO WATCH.

EVERYONE LOVED ADALIA, AS SHE WAS AN INSPIRATION TO THEM.

SADLY, ADALIA PASSED AWAY ON JANUARY 12, 2022, AT JUST 15 YEARS OLD BECAUSE OF PROGERIA. HER FAMILY AND FANS MOURNED HER LOSS, SHOWING HOW IMPORTANT SHE WAS TO THEM- AND EVEN THOUGH HER LIFE WAS SHORT, SHE LEFT A HUGE IMPACT ON EVERYONE.

ADALIA'S STORY REMINDS US TO BE BRAVE AND BE HAPPY FOR WHAT WE HAVE.

LINK<u>ADALIA ROSE</u>

Tiffany & Linda Wedekind- The Progeria Legends

TIFFANY WEDEKIND IS A 45 YEAR OLD WOMAN WITH PROGERIA. SHE IS THE 2ND OLDEST KNOWN PERSON WITH PROGERIA. SHE WAS DIAGNOSED WITH IT IN HER LATE 20'S WHICH IS WHY SHE HAS LIVED SO LONG. IF A PERSON HAS PROGERIA, THEY ARE USUALLY DIAGNOSED WITH IT WHEN THEY ARE NEWBORN OR VERY YOUNG.

SHE IS LIVING IN THE BODY OF AN 83-YEAR OLD WOMAN WHEN SHE IS ACTUALLY IN HER LATE 40'S.

LINKTIFFANY WEDEKIND PROGERIA LINKTIFFANY WEDEKIND PROGERIA



Tiffany & Linda Wedekind- The Progeria Legends

TIFFANY WEDEKIND LOVES AND DOES LOTS OF YOGA WHICH HELPS HERE STAY FLEXIBLE AND HEALTHY AND HELPS HER MAINTAIN A HEALTHY IMMUNE SYSTEM. SHE IS APPROXIMATELY 4 FEET, 5 INCHES TALL AND WEIGHS AROUND 68 POUNDS WHICH IS AROUND 31 KILOS. SHE LOVES DANCING, TRAVELING, AND HANGING OUT WITH HER FRIENDS. SHE LIVES IN COLUMBUS, OHIO. HER BROTHER CHAD ALSO HAD PROGERIA, BUT HE SADLY PASSED AWAY. SHE HAS 2 BUISENESSES. SHE RECYCLES BEER BOTTLES INTO CANDLES AND THE OTHER ONE IS TO CLEAN UP OCEANS.

TIFFANY'S MOM ALSO HAS PROGERIA, BUT ITS SO MILD THAT SHE BARELY HAS IT, SO SHE IS *TECHNICALLY* THE OLDEST LIVING PERSON WITH PROGERIA, BUT ITS SO MILD THAT IT DOES NOT COUNT AS MUCH AS HER DAUGHTER. HER NAME IS LINDA AND SHE IS A RETIRED NURSE AND IS 73 YEARS OLD.

TIFFANY AND LINDA WEDEKIND

Did you know?

• Progeria affects 1 in 20 million people in the whole world?

Conclusion

In conclusion, progeria is a disorder that is caused by the genetic mutation in the lamin a gene. The mutation causes lamins to turn into progerins. The progerins are not detected by proteins like c-gas and STING which activate your immune response. Progeria usually affects children that are around 2-3 months old. The symptoms start to clearly show at around 1 year old.

Some symptoms of progeria include: a large head, protruding eyes, visible veins around the eye area, a high-pitched voice, hair loss (including eyelashes and eyebrows), etc.

There are no known cures for progeria, but there are some treatments. Treatments for progeria include; physiotherapy, eye vision therapy, and hearing aids for tight joints, eye, and hearing loss. A nightly dose of low-dose asprin can also reduce the risk of heart attacks and strokes. The only known actual treatment for progeria is lonafarnib, which is sold under the brand name Zokinvy.

Progeria can also be treated with calcitriol, a type of vitamin D that rescues more cells from the glitches caused by progerin, making you gain more normal cells than progeria cells over a long period of time.

Some important people with progeria that I would like to mention are Adalia Rose and Tiffany Wedekind.

Adalia Rose was a famous YouTuber with progeria. She was born on D ecember10, 2006, in Texas and she was diagnosed with progeria as a 3-month old baby. She sadly passed away on January 12th, 2022.

Tiffany Wedekind is the oldest person with progeria. This is because she was diagnosed with it in her late 20's instead of a baby.

I hope that you learned something new in this presentation and I hope that you liked it. Thank you for listening. You can now ask any questions.