FANTASTIC SIBLINGS

In the documentary given out on CHOPS SYNDROME, featuring Leta and Mario,

scientist Dr Neil Hackett remarked

“the children with CHOPS are charming,

they are great kids

and the siblings especially are fantastic,

helping the CHOPS children out.

In this newsletter we want to bring praise to all the siblings.

WE APPLAUD THEM.

Some of the siblings have the following to say:

Leonard Lynn

When living with a family member who can't exactly communicate within the expected conventions you tend to believe they're simply incapable of doing so. I think my brother's realized this.

You, presumably a family member or close associate, may believe you are immune to this fallacy; as I often presume I am.

Despite knowing him nearly my entire (conscious) life, I often find myself talking down to him, only to be snapped back into reality by his (classic) face of scorn.

It is incredibly difficult, not only to communicate, but also to find some sense of relative "normalcy" in your life; a term which is heavily subjective but I digress.

What I find helps both him to fully express himself, and I, to see his true personality, is a creative outlet. Whether it be music, baking, or especially videogames, it is surreal to be able to communicate fully using a different medium. I encourage you to find something both of you enjoy if you haven't already.

It is often very annoying trying to deal with him, an inability of me to understand what he wants, an inability of him to do what I want. Especially with him becoming a teenager, he's become… well more like a teenager, adding fuel to the fire.

I don't know the experiences of other people, and I am incredibly biased seeing how we had entirely grown up with each other. I am incapable of fully understanding what you are going through.

I hope that you've gained something from my experience, even if that is just another view point.

As I end off, I'd like to provide some advice. To the best of your ability, treat them like you would anyone else (I know, how original), but I firmly believe it'll help both of you in the long run.

Thanks for reading-

Leonard Lynn (Brother of Edward, Teddy, Lynn).

OIHAN

My experience as the sibling of someone with CHOPS syndrome

Since I was little, one of the hardest things has been watching my sister try to play with other children, only to be rejected. Sometimes even the parents would tell their kids not to play with her. That's a memory that stuck with me and still hurts. Even now, when we walk down the street, people, often children stare at her as if she were something strange, as if they couldn't understand she's just different. Those looks, though silent, cut deep.

Now that my sister is 18, the most painful part is seeing how life keeps moving forward for everyone else, but not in the same way for her. Because of her condition, she remains both physically and mentally in a more childlike stage. Still, there are moments when she shows signs of maturity, she's aware she's growing older, and that makes her want to do things people her age usually do: have friends, enjoy some independence… and seeing that many of those things are out of reach is heartbreaking, for her and for us as a family.

Personally, it's been very hard to witness all the times she's been hospitalized and had surgery. I was just a teenager, and each of those moments forced me to grow up quickly. Watching someone you love suffer, changes you. It makes you appreciate health, family, and those little everyday moments most people take for granted.

To other siblings going through something similar, I would say: try to understand your brother's or sister's situation, even when it's hard but also try to understand what your parents are going through. This journey has likely been incredibly tough for them, and it's not over. Support your sibling as much as you can, always with their well-being in mind. But also know that sometimes, stepping back is okay. There may be moments when you don't know how to handle it emotionally, and that's valid. We must also remember that we have the right to live our own lives, as much as possible, without guilt.

SPANISH VERSION ON NEXT PAGE

Mi experiencia como hermano de una persona con síndrome CHOPS

Desde pequeño, uno de los momentos más duros ha sido ver cómo mi hermana intentaba jugar con otros niños y estos la rechazaban. A veces incluso los propios padres les decían que no jugaran con ella. Es una imagen que se me quedó grabada y que aún duele. Todavía hoy, cuando vamos por la calle, hay personas —incluidos niños— que se le quedan mirando como si fuera un ser extraño, como si no entendieran que simplemente es diferente. Esas miradas, aunque parezcan insignificantes, hieren.

Ahora que mi hermana tiene 18 años, lo más difícil está siendo ver cómo la vida avanza para todos, pero para ella no de la misma forma. Por su enfermedad, sigue atrapada tanto física como mentalmente en una etapa más infantil. A pesar de eso, hay momentos en los que demuestra que ha madurado, que es consciente de su edad, y eso le despierta el deseo de hacer cosas propias de una persona de su edad: tener amigas, más independencia… y darse cuenta de que muchas de esas cosas no están a su alcance es muy doloroso, para ella y también para nosotros como familia.

Personalmente, ha sido muy duro vivir todas sus hospitalizaciones y operaciones. Yo era aún un adolescente, y cada una de esas experiencias me obligó a crecer de golpe. Ver sufrir a alguien a quien quieres tanto te cambia, te hace valorar la salud, la familia y los pequeños momentos cotidianos que a veces damos por sentados.

A otros hermanos o hermanas que estén viviendo algo similar, solo puedo decirles que, aunque a veces cueste, intenten comprender la situación de su hermana o hermano, pero también la de sus padres, que seguramente lo han pasado muy mal durante todo este proceso, y aún tienen un largo camino por delante. Es importante intentar ayudar en lo posible, siempre teniendo presente el bienestar del hermano. Pero también hay que entender que, en ciertos momentos, tomar distancia puede ser necesario. No siempre sabemos cómo gestionar lo que sentimos, y está bien. Nosotros también tenemos derecho a vivir nuestras propias vidas, en la medida de lo posible, sin culpa.

Kyah, 6 years old says:

Zarli is my favourite star.

Kade, 11 years old:

Zarli is difficult to manage but she has beautiful parts about her. I love that she can walk and how she communicates.

CONGRATULATIONS TO JAMES AND EDS WITH THE BIRTH OF THEIR BEAUTIFUL TWINS

SIBLINGS TO THE LATE ANGEL ZIIA

After a heartbreaking loss of our dear Ziia, we've been blessed with a new light in our lives. We've had our share of heartache, but now we are filled with hope and anticipation for the future. Meet our miracle babies

Tania and Daniella

A reminder that even in darkness, love can bring forth new beginnings."

James and Eds

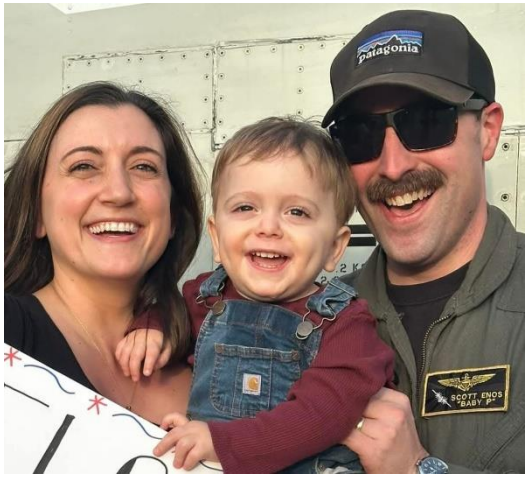
Antonio, born 5 May 2025.

Mario, admiring his baby brother.

NEWCOMMERS TO THE CHOPS SYNDROME DIAGNOSED GROUP

Ollyver Hopkins-Dukes, born

December 25, 2020 at 30 weeks



Left: Scotty Enos and parents from US.

Çağan Bozkurt, 11 years old from Turkiye, recently diagnosed.

IN AN EFFORT FROM SOUTH AFRICA TO RAISE AWARENESS ON CHOPS SYNDROME AND ASSIST WITH FUNDRAISING, 500 IZZY DOLLS WERE MADE AND SENT TO FONDAZIONE CHOPS IN CALABRIA.

Sophia, with love for Salvador and CHOPS syndrome research

KnitKnit (Suzanne) & Salvador with love to FONDAZIONNE CHOPS

Marieta with love for Fondazione CHOPS pediatric research

Made with for FONDAZIONNE CHOPS

Granny and friends of Salvador to raise awareness on AFF4 gene anomaly

Made uniquely for FONDAZIONNE CHOPS Lyn

Marieta&Magda, with for Fondazione CHOPS,

Lyn, Michelle and Patty

Anita & Carla made these with love towards pediatric research piloted by FONDAZIONNE CHOPS

Elmarie& Deborah from Springbok to FONDAZIONNE CHOPS

Granny from Gordons Bay, SA

Knitted with much love towards FONDAZIONNE CHOPS to do pediatric research Sanet

FONDAZIONE CHOPS ETS RESEARCH

Since May 2023 the CHOPS Rare Disease Foundation has funded 5 research projects:

*3 basic research projects, including one $200,000 and two seed grants of $20,000

*1 applied research project focused on pharmacological repositioning and conducted with the company Unravel Bioscience Inc.

*1 research project at Children's Hospital in Philadelphia to test drugs Identified by Unravel on cellular lines from patients suffering from CHOPS.

Many thanks for your support.

Keep supporting us please: fondazionechopsets.com

CHOPS SYNDROME

CHOPS SYNDROME is on the AFF4 gene anomaly, ONLY on the following very specific window.

253 – 260

It was researched from 2012 – 2015 on three patients at the Philadelphia Children's Hospital by drs Izumi and Kranz. CHOPS acronym can be laid out as follow –

Cognitive delay & coarse features

Heart defects

Obesity

Pulmonary issues

Short stature

Patients also have hearing problems, ocular complications and there are only 3 in the group of 36 who has more than 50 words vocabulary. The others are non-verbal.

Crawling and walking are severely delayed and they have a low muscle tone.

UNDERSTANDING THE DIAGNOSIS OF CHOPS SYNDROME

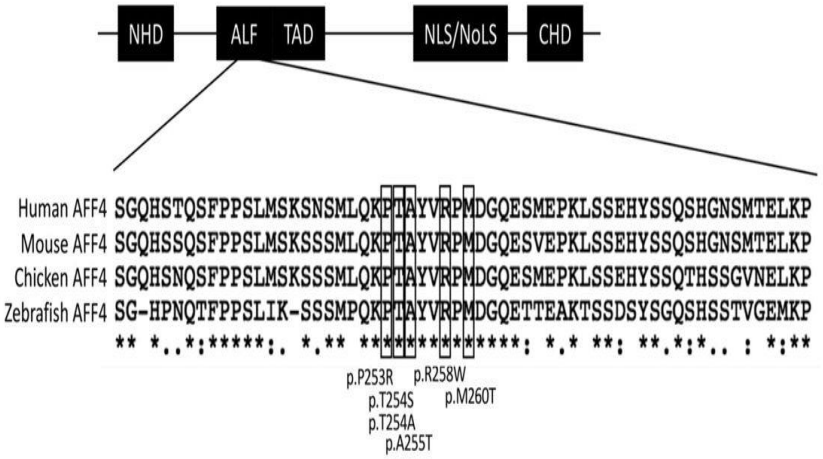


FIGURE 5 Locations of *AFF4* mutations identified in individuals with CHOPS syndrome. All of the missense mutations were de novo, and mapped to the evolutionally highly conserved ALF homology domain of *AFF4*. NHD, N-terminal homology domain; TAD, transactivation domain; NLS, nuclear localization signal; NoLS, nucleolar localization signal; CHD, C-terminal homology domain. Human AFF4 protein sequence is NP_055238.1, mouse AFF4 protein sequence is NP_291043.1, chicken AFF4 protein sequence is XP_015149549.1, and zebrafish AFF4 sequence is XP_005173956.1

CHOPS TASK FORCE

FAMILY GROUP

This group was selected by Fondazione CHOPS ETS in 2024

Dani McLennan, mother of Jakob from Australia

Griselda Franco, mother of Francisco from US

Jennifer Rayman, mother of Amellia from Canada

Josue Susperregi, father of Eider from Spain

Kayla Caceres, mother of Kyrie from US

Lainey Moseley , mother of Leta from US

Lisa Mcintosh-Conlon, mother of Mae from UK

Manuela Mallamaci, mother of Mario from Italy

Rae-Dawn Sandilands, mother of Salvador in South Africa

Suzanne Steyn, grandmother of Salvador (Secretary)

The main role of the Task Force Family group will be to share collected ideas raised during the meetings with the Medical board.

THERAPEUTIC ASSISTANCE FOR CHILDREN AFFECTED BY CHOPS SYNDROME

Rinnie Skapp, mother of Zarli who is 9 years old and could not walk till recently and is also non-verbal, writes –

The ongoing battles not only I have for Zarli, so many families are enduring these battles ontop of the care we provide for our loved ones. The benefits Zarli's therapies have had are amazing, and I will continue all I can to provide these opportunities for her.

ABC Capricornia · **Follow**
12h ·

Rinnie Skopp's daughter Zarli took her first steps thanks to an intensive child therapy service earlier this year.

But after the momentous milestone NDIS claimed the program was not worth the money.

Get the news that matters to you straight from the source. Download the ABC NEWS app: https://ab.co/abcnewsapp

abc.net.au
As Zarli, 9, took her first steps at therapy her NDIS funding was cut

After intense therapeutic sessions Zarli proudly shows off her ability to walk, even when she needs the wall to give her stability.

I did it!!!!

🧬🔬Three scientists for CHOPS

We present them to you, they are the winning researchers of our research call for 2024 according to the evaluation of our medical scientific committee.

🧒🔬 Kosuke Izumi, researcher at the University of Texas Southwestern Medical Center in Dallas, first in grading for the contribution of 100 thousand + 100 thousand dollars (about 92,500 + 92,500 euros),
Researcher with Ian Krantz of CHOPS syndrome first diagnosed at Children's Hospital Philadelphia in 2015.

👩🔬 Ilaria Parenti researcher at the Institute of Human Genetics at the Essen University Hospital of Duisburg University in Germany, in second place with a contribution of 20 thousand dollars (about 18,500 euros).

🧒🔬 Jun Young Sonn researcher at the Baylor College of Medicine in Houston in Texas, equal merit with Dr. Parenti, for an equal contribution of 20 thousand dollars (about 18,500 euros).

Between Europe and the United States, at very prestigious research bodies, these young and brilliant scientists will carry out basic research on CHOPS syndrome, with the aim of clarifying its biological mechanisms.

Aspera ad astra to them, but above all to our children ✨☆✨

And we'll never get tired of saying that all of this was possible because of your donations, you're part of this big dream. ♥



🎶 An evening that touched the heart.
Heartfelt thanks to Christus Vincit, to Maestro Ripepi, to the extraordinary voice of Sherrita Duran, to all the choirs, the choir of the diocese of Rome and all those who came from all parts of Calabria, to the talented singer Cecilia La Rosa, to all the staff, to the orchestra, to Don Francesco Cristofaro and to everyone who contributed to making this concert an unforgettable event.

Music brought together emotions, solidarity and hope. Together, we've made an important step towards the future of CHOPS Syndrome research.
💙 Thank you to those who chose to be there, to support and to believe.

🎉 The value of a business is also measured by its social impact.

Any business or freelancer that decides to support the CHOPS Rare Disease Foundation makes a choice that goes beyond business.

💙 Supporting CHOPS syndrome and rare disease research means:

🔷 Investing in Science
🔷 Supporting families facing daily challenges
🔷 Strengthening your company's social responsibility
🔷 Access to tax advantages
🔷 Communicate your ethical commitment

🤝 Join us. Become part of our mission.

SIMPOSIUM PLANNED

FOR SUMMER OF 2026 IN ITALY

WE AIM TO EXPAND RESEARCH

A cure for CHOPS SYNDROME is possible!

We need much more funds to enable further research to make it happen.

PARENTS, FAMILY, FRIENDS &

FUNDING FOUNDATIONS

PLEASE START ASSISTING

Regular monthly donations will be of great help!!

Una tantum = once off

Mensile = monthly

Annuale = annually

https://fondazionechopsets.com/en/donate-now/