Picture taken during CHOPS SYMPOSIUM IN PHILADELPHIA, JULY 2024

NEWSLETTER IN HONOUR OF ALL THE CHOPS PARENTS

Leta and Lainey

MAY THE FESTIVE SEASON BRING PEACE, JOY, LOVE AND REFRESHING FAMILY TIME TO ALL

FUNDRAISING SUCCESS

A gigantic effort of fundraising was once again done by parents, family and friends of SUPER MARIO.

The CHOPS family wants to give the biggest appreciation and applause to them all one again.

This enabled Fondazione CHOPS Malattie Rare to commence with the follow-up stage of research.

THREE NEW RESEARCH PROJECTS SIGNED AND PAID FOR BY FONDAZIONE CHOPS ETS

Start the search for the BASE for the CHOPS 🚀

We are thrilled, enthusiastic, proud and above all hopeful to announce the official launch of 3 international projects with prestigious research bodies and researchers to identify the biological mechanisms that characterize CHOPS syndrome, an ultra-rare, multi-organ genetic disease and neurodevelopmental.

All this THANKS to your donations!!!!!
THANK YOU to the invaluable support of our medical-scientific committee led by **Del Pinto e Associati - Studio Legale** THANK YOU to the tireless team of **Ian Krantz** 💔💔💔

Full details in the article of **LaC News24**
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https://www.lacnews24.it/sanita/il-piccolo-mario-affetto-da-una-malattia-rarissima-la-fondazione-gestita-dalla-mamma-avvia-tre-nuovi-progetti-di-ricerca-ymmlrtrp

CHOPS Rare Diseases Foundation launches investigation into biologic...
Three international contracts signed
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https://www.einpresswire.com/article/766778131/chops-rare-diseases-foundation-launches-investigation-into-biological-mechanisms-of-the-rare-syndrome 14:09

PLEASE FOLLOW THE DIFFERENT GIVEN LINKS FOR MORE INFORMATION

RESEARCH PROJECTS APPROVED BY

THE MEDICAL ADVISORY BOARD OF FONDAZIONE CHOPS ETS

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SNIPPETS FROM ANOTHER PRESS RELEASE

Three international contracts signed

ITALY, December 6, 2024 /EINPresswire.com/ -- "We have signed 3 contracts and are now ready to see basic research investigating the as-yet-unknown biological mechanisms that characterise CHOPS syndrome come to life. These are important contracts with authoritative institutions. Kosuke Izumi, Ilaria Parenti and Jun Young Sonn. They are the professionals who mark a crucial moment in the Foundation's work". Manuela Mallamaci, president of CHOPS Rare Diseases Foundation and the mother of little Mario, announced this.

The CHOPS Foundation is the world's first and only non-profit organisation to research a cure for this rare, multi-organ syndrome.

"We do not intend to stop, but we need the solidarity and support of the people. The Foundation has already started projects with more than 300,000 euros raised. Complex and challenging projects. It will be crucial,' Mallamaci emphasises, 'to continue by activating the best minds on an international level to shed light on this disease. So, it will be necessary to continue raising funds. Path Along, which we are also using, is the CHOPS documentary donated and created by the director Antonio Melasi.

The Foundation is committed to promoting its work internationally and involving families in this awareness-raising process.

EMOTIONS AND COPING STRATEGIES OF PARENTS

Jennifer Rayman writes -

Grieving my idea of a perfect child.

Before she was born, I imagined all the fun things we would do and the books we would read. How she would look and what she would grow up to be. I had this made-up prefect child in my mind, but reality came in the form of a CHOPS diagnosis.

Since it took years to eventually get a diagnosis, I lived day to day in the real world of an atypical child. Navigating the missed milestones, endless tests and hospital visits, and the worst of all – questions and looks from other parents at the playground. I felt so sad and alone and, tremendously guilty for feeling that way. Then, I came across a great book that validated my feelings and helped me on my path to active acceptance.

The book is called **'Not What I Expected', by Rita Eichenstein PhD**. It explains how parents of a child with specials needs move in and out of the 5 stages of grief.

1. Denial/retreat
2. Anger/aggression
3. Bargaining/seeking control/seeking solutions
4. Depression/isolation/shame
5. Active acceptance/equanimity/integration

This book set me on my path to label my feelings and accept them. I understand that our normal is different from other people's normal. I can better celebrate our wins and not fall into a state of despair when we experience a set-back. I see how truly exceptional my child is and celebrate the fact that I have a best friend for life.

From Lainey Moseley -

Leta was born in 1997, weighing 5 lbs 4 oz (2,38 kg) … she ended up in the NICU for 3 weeks when they discovered a PDA and she needed heart surgery soon after. We didn't start to sense there were real problems until she didn't hit any of her 6 and 9 month milestones and she started having a lot of pneumonias. Her fist diagnosis was Cornelia de Lange Syndrome, then years later they looked at metabolic disorders but in the end we stopped caring what was wrong and just dealt with the NOW. Her official diagnosis didn't come until she was 18 years old. She was the first diagnosed with CHOPS Syndrome
We were thrilled when we received the CHOPS diagnoses because we learned that it was a de novo mutation and that meant it wasn't passed down to the other kids and I was also so relieved that it wasn't something I had done wrong while pregnant to cause her issues. We also knew that this was exciting scientifically and that we had a tremendous responsibility to find and help other families get diagnosed.

There was no support group to lead and assist us, only Leta's genetic counsellor at CHOP (Ian Krantz). There was no additional support. No financial support and/or grants for our government.
You go through the different stages of grief when you learn you have a child born with a disability - the best thing you can do is to move through denial, anger bargaining, depression and get to acceptance as quickly as you can because that is when you can be the best parent for these amazing kids.

The advice I can give to other parents in the CHOPS journey? It is so hard to see the silver lining at first, but I promise that these kids will create a special type of meaning in your life that no other child would ever be able to offer. They will be your teacher and inform so many of your life decisions all in the best of ways. This path will be hard and it is not the path you would have probably chosen, but you will grow so much and learn so much about yourself and other people … nothing truly special is ever easy and our CHOPS kids are truly special. They are bundles of unconditional love and how lucky are we to get to have all that love in our lives.

Kayla Caceres recalls -

Memories of the time from birth of Kyrie up to the time of diagnosis.

Mix of emotions, so elated and in love, seeing him as perfect but then worried and confused why so much was going wrong and why there were multiple problems.

There was hope that nothing was wrong except hearing loss because physically he appeared "normal" and would reach his milestones right at the end of the timeframe he was supposed to, but once he hit 8 months all the milestones were falling behind

Emotions on receiving the CHOPS diagnosis.

Worry, despair, hopeless, fear for his future and what it would be like, feeling punished, some relief knowing there was a diagnosis – took 2 years to get a diagnosis, "didn't want to belong to the special needs club", some denial

This probably lasted for about 2 weeks and then I accepted it. I had to properly grieve what I thought the future was going to be. Sometimes when new problems arise I fear and worry but am able to get back grounded more easily. Its helped me be more present and accept what is. I focus more on what we can do and control verses things being "bad"

Support group to lead and assist?

I have an individual therapist; read a booked called **Special by Melanie Dimmitt**; looking up CHOPS kids social media in secret. Took me some time to reach out and join the group. I was still having a hard time accepting it.

My therapist assisted me and I talked to other parents of special needs kids, the book really helped me

I am a therapist myself so I kind of understood what needed to happen emotionally to get through it

Since he was 2 months old and due to the hearing loss that was caught when he was 1 month old, we received assistance through our local school district. A special needs early development teacher would come to our house and teach us different ways to work on developmental skills

We received grants from our government for hearing aids.

Continues on next page

The school district provided services for general education regarding development, occupational therapy, physical therapy, and deaf and hard of hearing services. We met with each individual 1xs/week

Once he turned 3 years old, he was able to enroll in special needs preschool and then received services through our county for additional help like funding for diapers, respite care

We also qualify for in home support services where our caregivers get paid to help and the caregivers could be your family members or yourself

Emotions differ during the developmental stages.

Now I just enjoy the ride as best as I can. I realize things will happen when they do and if they don't there are supports for Kyrie. I am more hopeful for his future because he is thriving. When he was a baby, I just didn't know what our life would look like. And now that he is 6, he loves school, can do way more academically than I expected, and is happy. My thought was if Kyrie is happy with his life why do I have to be sad for it. That would just make him think there is something wrong. So I try to be more like him!

There are still hard days and we are in developmental stages for much longer, but I do things to take care of myself and we are lucky to have a lot of family support so I can handle him and any challenges better.

Advice to other parents in the CHOPS journey.

As cheesy as it sounds, Kyrie having CHOPS has made me have an even greater appreciation for life. I learned nothing is ever guaranteed and we are all just making the best of it.

Allow yourself to feel and think whatever comes up because you do need to grieve. But don't get stuck there. Find a good support system that will hear you out weather that's a therapist, pastor, family member, friend, online support group, a book.

I highly recommend the book **Special by Melanie Dimmitt**. It gives you the words to what you are experiencing. Sometimes you don't even know what you are thinking and feeling but the book helps with that. She just highlights all the ups and downs of having a child with special needs.

Get your kids in therapy services as soon as you can. It's a lot of work for them and us but totally worth it! Do all the things you would do with neurotypical kids – get outside, do family trips, enroll them in activities.

FONDAZIONE CHOPS ETS

RELENTLESS, NON-STOP FUNDRAISING IN ORDER TO ENSURE RESEARCH TO BRING HOPE TO CHOPS SYNDROME PEOPLE AND OTHER ANOMALIES

Fondazione CHOPS ETS
29 Nov ·

Join us for the evening ✨ SOLIDARITY FLAVORS' ✨

🍴 More than 40 chefs will accompany you on a real food itinerary, with extraordinary dishes to discover and taste.

🍷 40 wineries will guide you on a journey among the finest wines, for perfect pairings and unlimited tastings.

❤️ The proceeds will be donated to the Chops Foundation, to support research on an ultra-rare disease.

Check out all the participating restaurants and wineries!

📌Where: Exforo Restaurant, Prato della Valle, Padova

🗓️When: Sunday, December 22, 2024

Cost: €40 per person.

👉 Reserve your place now at the link bit.ly/Sapori_Chops

ITALY STANDING TOGETHER FOR CHOPS SYNDROME

UNDERSTANDING CHOPS SYNDROME

PRECISELY WHAT GENETISISTS NEED TO CONCENTRATE ON TO MAKE DIAGNOSIS

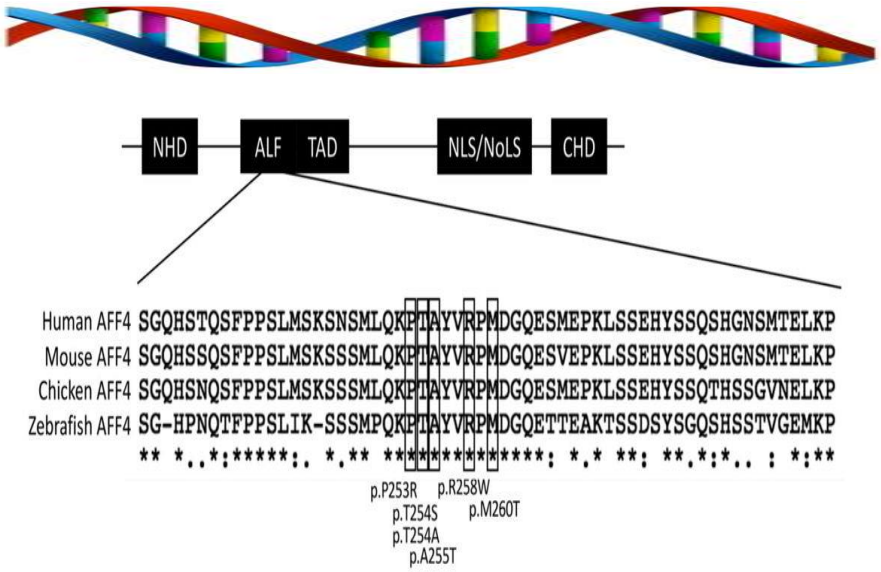

FIGURE 5 Locations of *AFF4* mutations identified in individuals with CHOPS syndrome. All of the missense mutations were de novo, and mapped to the evolutionally highly conserved ALF homology domain of *AFF4*. NHD, N-terminal homology domain; TAD, transactivation domain; NLS, nuclear localization signal; 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

SCREENING OF THE FOLLOWING FILM IS AVAILABLE AS FUNDRAISER FOR RESEARCH,

INITIATED BY FONDAZIONE CHOPS ETS.

Anyone who wishes to arrange a screening and contribute towards the paediatric research,

can contact Lainey Moseley at lwmoseley@comcast.net

Funds to be paid over to Fondazione CHOPS ETS - contact manuela.mallamaci@gmail.com

TOORARETOCARE DOCUMENTARY

www.tooraretocare.com

Rare diseases affect more people than Cancer and AIDS combined.

There are over 10,000 rare diseases in the world. 1 in 10 people worldwide have a rare disease and 50% of these rare diseases affect children. But unfortunately, there are treatments for less than 5% of those diseases. As such, 30% of these children don't make it to their 5th birthday. And while no one thinks their child will be born with a rare disease, it can happen to anyone – nature does not discriminate.

Few can appreciate the battle families of kids with rare diseases fight. **The mission of our film is to raise awareness about the diagnostic and cure odyssey of rare disease kids that falls primarily on the advocacy of parents.** We ask the question, why isn't more being done to save these kids? And if this was your child, what would you do?

When parents are told that their child has a rare genetic disease, they are advised that nothing can be done, that there are no treatments, and to "go home and enjoy your child." But the science and success of gene and cell therapy is changing the landscape for rare diseases and giving families hope.

Elizly writes –

Salvador's incredible journey developing communication

As a parent of a "bespoke" little boy his challenges became ours. When I started realising the things Salvador would be challenged with, I would imagine his life and what it would be like living with those challenges. To date I can and could accept and imagine him living a purposeful life with all these challenges, but I could not imagine what it would be like not to be able to fully communicate his thoughts, ideas and emotions. His challenge with speech and the future it could hold for him or WITHOLD from him absolutely broke my heart. Today, a year later I am filled with hope, admiration and pride for the miracle of speech and the angel that was placed on Salvador's path to aid and guide him into where he is today.

We have learned that early intervention in guidance with specialist developmental therapy and repetition has been tremendously successful with Salvador. We have had so many simple strategies with significantly positive outcomes for Salvador like making lists of the sequence of events for a morning routine or just getting dressed, fed and out the house.

We would like to share Kirsty's report on Salvador's first year in therapy and some of the strategies she has used to assist his development in the hope that this may help others.

We would also like to invite other speech therapists in our CHOPS FAMILIES to join in sharing ideas, strategies, learnings and successes. If you as a parent together with your therapist want to join this group, please contact Isabel Suzanne Steyn.

[issteyn@yahoo.com or SPEECH4CHOPS what's app group (0)82 889 2948]

The hope is that our shared experience and **learnings** will eventually become a guide for other chops parents and therapists.

See Dr Kirsty Bastable's report of Salvador's speech on page 13 - 15 ……

HOW TO APPROACH PROSPECTIVE FUNDERS/DONERS TO OUR RESEARCH PROJECT

Most countries have the GOFUNDME and GIVEALITTLE campaigns where one can raise awareness of the situation and request for people/organisations to donate.

For this research project, we would like these donations to be deposited directly into the bank account of Fondazione CHOPS ETS

(see 2nd column ITALY (EUROPE) in table below for banking details).

Should there be doners from USA who want tax rebates for the amount funded, the option is there to follow CHOPS foundation global, as indicated in the first column below.

Please ENSURE to also click the link indicating that the donation should go to CHOPS.

Request family and friends to make bequests towards Fondazione CHOPS ETS in their wills

FUNDRAISING FOR CHOPS SYNDROME RESEACH
Thank you for contributing towards a very important project
For more information on CHOPS, please visit
fondazionechopsets.com and/or www.chopssyndromeglobal.org

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Click on DONATE button, complete detail, specify it is for CHOPS, choose the monetary value you are donating in.	Name: Fondazione CHOPS Malattie Rare Ente del Terzo Settore	Gofundme.com IDENTIFY **ITALY BANKING DETAILS
	IBAN: IT81K0103016303000000974528	BackaBuddy (South Africa) IDENTIFY **ITALY BANKING DETAILS
	Other ways to donate: you can go to the fundraising page "Un aiuto per Mario e non solo" and donate via credit card or paypal.	
	BIC/SWIFT: PASCITM1RC2	

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Speech therapy Report

Name: Salvador Sandilands-Steyn Date: 1 December 2024
DOB: 10/07/2021 Age: 3 years 4 months
Diagnosis: CHOPS Syndrome ICD-10: Q87.8

No. Sessions: 37

Salvador began intervention with me in January of 2024. I had assessed him in November of 2023 and provided feedback to his family. The assessment indicated an approximately 9 month delay in Salva's expressive language as well as gross, fine and oral motor difficulties. Recommendations from Salva's assessment included the continued implementation of signing in combination with functional communication approach where different functions of communication are taught and combined e.g. starting an activity and who – I go. In addition oral motor intervention and training for a behavioral hearing assessment were recommended.

Goals, Intervention and Results:

Oral Motor Skills development:
Goals with Salva's oral motor skills have been the development of his tongue, lip, cheek and breathing control and strength such that they are at a level which is sufficient to support his speech.

Intervention with oral motor skills has included play with whistles, bubbles and a mirror.

Salva has shown lovely development of his oral motor skills. From only being able to blow a bubble off the bubble wand with his mouth in a relaxed open position he is now able to blow all the whistles in my collection more than 10 times in a row. In addition we are now using the whistles to work on listening skills, having fun blowing different rhythms and strengths. Salva is learning how to listen and copy rhythms although he finds these difficult if they are more than 4 blows long. We have also recently added movement to our whistle play – walking while blowing, as speech is not something that we only do when seated. This has been difficult for Salva, but he did manage to blow while walking.

Receptive and Expressive Language:
Goals for Salva's receptive and expressive language have included the development of his vocabulary, increases in utterance length and the understanding and use of correct grammar, including plurals, questions, 3 word sentences, pronouns, prepositions, negatives, co-ordinators (and), and the ability to construct a cohesive narrative.

Salva's language has been targeted through play, singing, reading stories, and constructing sequence stories.

Salva has made great progress in his language this year. He began the year using Makaton and producing occasional single words, and he has ended the year as producing up to 3 word sentences verbally. As Salva's language has progressed I have moved from intervention using both English and Afrikaans to intervention primarily in

English, with Afrikaans used only if needed for Salva's comprehension. However his family report that his Afrikaans speech and language development is progressing in line with his English. Salva's receptive language continues to progress ahead of his expressive language. Although his sentences are short, Salva is able to produce a narrative about stories, explaining what is happening, how and why. He is able to use pronouns, plurals, prepositions, adjectives and adverbs appropriately and spontaneously. I do feel that the length of sentences produced by Salva is limited both by his ability to produce speech as well as language difficulties. In spite of this Salva continues to try his best to express himself. At school recently Salva has presented with some behavioral challenges when he is tired, which may well be related to the effort it takes to express himself.

Speech Production:
Goals for Salva's speech production began with producing any speech and are now focused on producing individual speech sounds within running speech. Salva is able to produce most the sounds required for English in isolation or in initial position in a cv or vc syllable, including /l/, /r/, /sh/, and /ch/ (these are advanced for his age). However as he needs to produce multisyllabic words and longer sentences his sounds often begin to distort due to his oral dyspraxia. Due to Salva's age his self-monitoring of speech is not great, but we are working towards his being able to self-monitor and self-correct his speech as he goes. With longer sentences I have noticed that Salva at times presents with the phonological process of fronting of back (velar) sounds primarily /k/ and /g/ become /t/ or /d/. This is only when the sounds are at the end of a cvc production, and so I am not sure that this is a true phonological process but may be linked to oral motor difficulties. We will continue to work on this.

Swallowing and feeding:
Goals for Salva's feeding have included the development of the ability to bite, chew and swallow safely, and drinking using a cup. Linked to each of these was also swallowing while maintaining his head in an upright position – not throwing his head back, and not using his hands to push food into and around his mouth.

Feeding intervention with Salva has consisted of providing guidance to Portia and his mom's with regards to how feeding can be done in a manner which encourages the development of this. e.g. lateralised feeding, lateralised biting, licking off of an external surface.

A modified barium swallow was conducted with Salva when he was in hospital for management of his constipation. At the time I was most concerned that there may be nasal regurgitation of liquids or food. The swallow however showed that Salva has an excellent swallow. No nasal regurgitation was noted, not aspiration risk was evident and his swallow was fast and effective with no oral residue remaining.

Salva has made lovely progress with his feeding although he does still sometimes revert to using his hands to help him move food backwards in his mouth when he gets tired. He is able to chew and swallow all textures of food and is drinking well from a cup.

Summary and Recommendations:
Working with Salvador is such fun! He is an absolute charmer who really does try very hard. He has made fantastic progress this year, moving to becoming fully verbal,

developing his language and speech and progressing in his feeding. I have also seen lovely cognitive and attentional development as we have worked through the year.

During therapy Salvador does sometimes appear to have a short attention span, however he does also use this as an "excuse" when something is hard for him. I have found that giving him breaks between activities, and being clear about my expectations for the session for the most part manages this behavior effectively. For each session I set out the activities I would like to do in a row and we then work through them one by one. This appears to assist Salva in managing his impulsiveness as he knows what is coming next. He is also able to work through less appealing options while waiting for his favorite activities.

I am excited to see Salva continue to progress in the future.

Please do not hesitate to contact me if you have any questions.

Yours Sincerely

Bastable

Dr. Kirsty Bastable

EVERY BIT OF ASSISTANCE AND HOPE TO GET OUR KIDS COMMUNICATING IS OF INFINITE TREASURE

Please share your child's communication path with us in order to assist others.

AIMS FOR THE NEXT TWO YEARS

EXPAND RESEARCH - in joint venture with

Unravel Bioscience

Development of new therapeutics for complex disorders with unprecedented efficiency by integrating AI systems biology computation with rapid *in vivo* screening and clinical validation of discovered targets.

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THREE NEW RESEARCH PROJECTS

Financed by Fondazione CHOPS ETS

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

FUNDRAISING – we ALL need to put in an effort to get more doners to fill the monetary gaps.

MAKE IT BETTER FOR EACH AND EVERY ONE

OF OUR CHILDREN & FAMILIES