PHILADELPHIA SYMPOSIUM 2024

The families who joined the Symposium bonded and received fantastic news from scientists

THERE IS HOPE FOR A CURE FOR CHOPS SYNDROME!!

Attending families (front row from left) – Luce, Luigy (Italy), Leta (US), Amelia (Canada), Madison (US), Jakob (Australia), Eider (France), Andrew (US), Kyrie (US), Teddy (US), Mark (Philippines), Francisco (US) also attending and not on photo Liam (US)

Attending on line – Mae (UK), Mario (Italy), Salvador (South Africa)

OUR CHOP FAMILY HAD GROWN SINCE 2019

Kids diagnosed between (when CHOPS syndrome was found since 2015) from left:

Liam

Teddy

Aspen

Leta

Andrew

Jakob

Dr Krantz

Taylor

Madison

FEEDBACK FROM SYMPOSIUM

Manuela Mallamaci, mother of Mario and co-organiser of the second Symposium and founder of **Fondazione Chops ETS** writes -

2nd CHOPS Symposium 🌎🧬👩‍⚕️👨 '- The international symposium on CHOPS syndrome at the Children's Hospital of Philadelphia has concluded.

19 and 20 July: Two intense, very exciting days and of great scientific relevance.

Some numbers: 15 families from all over the world, 17 oral interventions by doctors, researchers and experts, with a total of 70 participants, even remotely connected. A huge success by all.

This symposium is the result of a great job and comes as a milestone at the conclusion of an intense year of activity.

We thank all the scientists and their families, we thank the hospital for hosting us, **Cool Cars for Kids** and CHOPS Syndrome Global.
We would like to thank Lainey Moseley, Sarah Raible, **Ian Krantz** and our president **Manuela Mallamaci** who material organizers of the event.

Made possible by

SCIENTIFIC COMMITTEE

Dr Ian D Krantz MD

Ms Sarah Raible CGC

ORGANIZING COMMITTEE

Dr Ian D Krantz MD

Lainey Moseley

Sarah Raible CGC

Manuela Mallamaci PhD

SPONSORS

Children's Hospital of Philadelphia

COOL CARS FOR KIDS

FONDAZIONE CHOPS MALATTIE RARE

CHOPS SYNDROME GLOBAL

FROM CHOPS FAMILY

INFORMATION THAT SYMPOSIUM MADE AVAILABLE

(written by Manuela Mallamaci)

The first day was dedicated to the contributions of specialist doctors and to personalized consultations between them and their families.
It has been described how CHOPS affects the normal development of various organs in the body. For example, children with CHOPS are often born with structural heart defects that most often require surgery. Even the intestine does not develop properly contributing to the chronic constipation that is observed in CHOPS. Furthermore, due to the limited verbal abilities found in children with CHOPS, it has emerged that alternative augmented communication methods may be essential.

In the afternoon of the first day many participating families, as well as holding out interviews with specialists, joined blood draws and skin biopsies, performed personally by Dr. Ian Krantz. The collection of these samples is being promoted by the Foundation with the intention of creating a biobank that is available to the whole scientific community for the basic study and development of potential therapies for CHOPS!

The second day was dedicated to molecular understanding of CHOPS and perspectives in terms of finding a cure. The Dr. also present with an oral contribution. Izumi the explorer together with Krantz of the syndrome. Izumi summarized her discovery, emphasizing how typical genetic mutation of CHOPS causes an abnormal accumulation of a specific protein that plays a key role in genetic expression.
Also present is Unravel, a company funded by us for the pharmaceutical screening project, which presented the results conducted by the extraction of 24 samples of patients suffering from CHOPS.

In the afternoon, the documentary film "RARE" produced by Lainey Moseley, Leta's mother was screened.
https://www.tooraretocare.com
This movie made us very emotional and at the same time further recharged and filled with hope.

TOORARETOCARE DOCUMENTARY

www.tooraretocare.com

UNDERSTANDING CHOPS SYNDROME

PRECISELY WHAT GENETISISTS NEED TO CONCENTRATE ON TO MAKE DIAGNOSIS

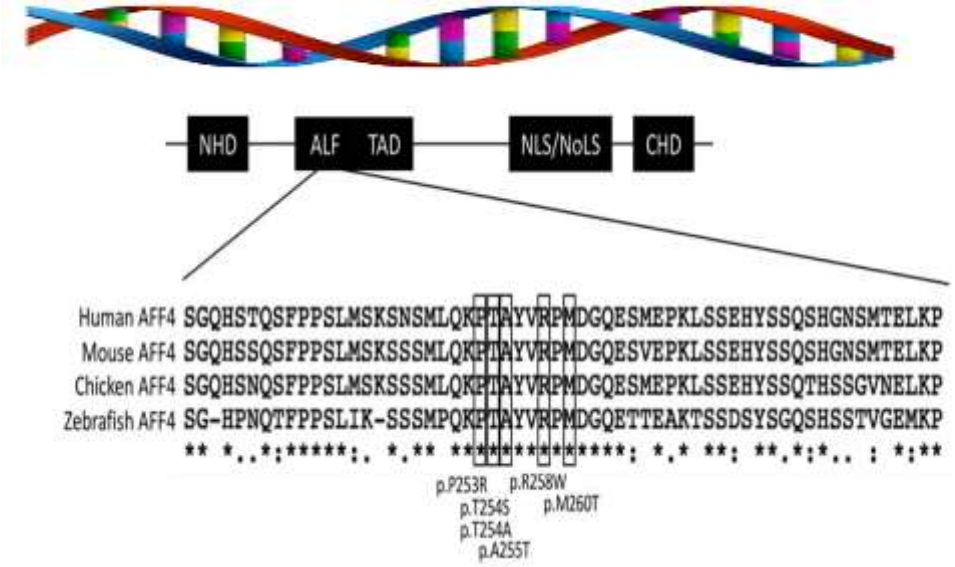

FIGURE 5 Locations of *AFF4* mutations identified in individuals with CHOPS syndrome. All of the missense mutations were de novo, and mapped to the evolutionally highly conserved ALF homology domain of AFF4. NHD, N-terminal homology domain; TAD, transactivation domain; NLS, nuclear localization signal;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

Medical and Scientific Advisory Board of Fondazione CHOPS agrees about focusing potential therapeutics searches on classical CHOPS patients, i.e. patients who have mutations/deletions in the domain described above.

Leta is doing a great job at the symposium as 'secretary' and ensures that all arrangements go according to plan.

FUNDRAISERS' ENTHUSIASM

Fondazione CHOPS ETS paid for the initial Unravel Bioscience research, co-funder the Symposium and funded research on speech and more ….

A few days post the symposium and fundraising continues with renewed enthusiasm.

Kayla started the fundraising a few years ago. A substantial amount is already available towards CHOPS syndrome awareness.

This isn't something that comes easy to us, but for this we need to put our pride aside and ask for your help. We need to raise as much money as we can so we can find a cure or at the very least improve the quality of life for our Mae and other children like her with CHOPS syndrome. Please, please share far and wide.

gofundme.com

Donate to Help Mae raise the money to find a cure for CHOPS syndrome., organized by Lisa Conlon

Spreading awareness and fundraising for

CHOPS in Cape Town.

Elizly Steyn, Salvador, Rae-Dawn Sandilands.

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

USA	ITALY (EUROPE)	OTHER COUNTRIES
CHOPS FOUNTATION GLOBAL	FONDAZIONE CHOPS ETS	
ive.rarevillage.org Research body To donate, go to chopssyndromeglobal	**It is possible to donate via bank transfer to:	givealittle.co.zn IDENTIFY **ITALY BANKING DETAILS
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 IBAN: IT81K0103016303000000974528 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	Gofundme.com IDENTIFY **ITALY BANKING DETAILS BackaBuddy (South Africa) IDENTIFY **ITALY BANKING DETAILS

Calling all CHOPS Warriors!

We are all fighting for something bigger than ourselves, and that is our kids. So that makes warriors of all of you in this disability journey.

But that also comes with an obligation to the next generation of families whose kids are born with the Aff4 gene mutation to move the needle closer to treatments and perhaps someday a cure.

Our CHOPS community is not large but as the founding members of a syndrome that was only identified 10 years ago, we have a responsibility to our kids and to science to help advance our understanding of CHOPS Syndrome and we also have a responsibility to help raise awareness in our communities and with the medical world that this syndrome deserves more research.

Genomic medicine has advanced so much in the past two decades. ultra rare single gene mutations like CHOPS Syndrome can be diagnosed within months or years of birth through exome sequencing and that earlier diagnosis means families are not just being satisfied with a diagnosis they are expecting and demanding treatment options.

Where once we were sent home and told "Good Luck", families are on cure odyssey's for their kids rare diseases.

The technology exists , gene therapies, ASO's, and some repurposed drugs, that can offer meaningful treatments for some of the rarest diseases.

The only problem is that the cost of developing these treatments for 95% of ultra rare diseases is still extremely expensive.

In 2024, we are now a community of 40 families and we have to continue the mission to do more than just gather community, we need to advance the scientific understanding of what causes CHOPS Syndrome, what the mechanisms of the Aff4 mutation but this takes money.

So please ask yourselves, what more can I do to help our community of CHOPS families?

Sadly, as an ultra-rare disease, no one else is going to care enough to do this work for our kids.

You all are the advocates for our CHOPS kids futures and we are only stronger together if we work together. If you would like help with ideas on fundraising in your community, please contact Manuela, Kayla, or Lainey.

manuela.mallamaci@gmail.com krcaceres@gmail.com lwmoseley@comcast.net

BONDING AND MAKING NEW CONNECTIONS

Teddy and Eider, Luigi and Teddy
Eider and Maddie, Andrew and Dad,
Luigy and Leta, Francisco with Mom and Liam,
Group photo with Mario and Salvador on line in background,
Eider and Maddie

No further analyses to be done at present.

The Medical and Advisory Board of Fondazione Chops ETS is discussing the optimal strategy which can include both in vitro and in vivo tests.

FAMILY TIES

Left to right:
Top row – Leta, Andrew, Jakob
Middle row – Madison, Francisco, Eider
Bottom row – Kyrie, Teddy, Luigi

Amelia
Mark
Luce

DR KRANTZ, receiving a gift from Leta, (representing the chops family).

Dr Krantz making time to meet our kids.

A gift to Manuela Mallamaci

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.

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