CHOPS SYNDROME

NEWSLETTER

THIS NEWSLETTER GIVES HONOUR TO KUZEY

FROM TURKEYE

Kuzey sadly passed away late April at the

tender age of 18 months only.

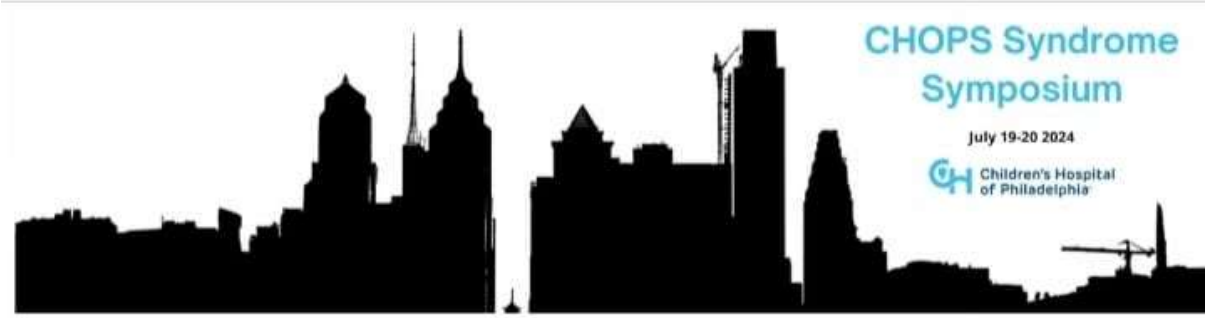
From his mother, Miraç - (translated)

Kuzey was a very strong angel, he resisted life very well, I think that an adult person cannot bear what Kuzey went through, but Kuzey resisted life many times. Even if his body leaves us, his spirit is always with us, I feel his presence every second, I miss him very much and I love kuzey very much❤️❤️

Our hearts and condolences are with the bereaved family. ♥

PHILADELPHIA SYMPOSIUM

The long awaiting second symposium is only one month away!



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

ALSO VISIT THE WEBSITE AT

chopssymposium.sciencesconf.org

Extracts form the website hereunder

OVERVIEW

The CHOPS Syndrome is an ultra-rare and multi-organ neurodevelopmental genetic disease. It involves several abnormalities that are present from birth. The name CHOPS is the acronym of Cognitive impairment and Coarse facies, Heart defects, Obesity, Pulmonary involvement, Short stature and Skeletal dysplasia, or the list of the main characteristics of the syndrome

It is caused by mutations at the AFF4 gene, which provides instructions for building a protein complex known as a super elongation complex (SEC). During embryogenesis, the ECS is involved in a process called transcription, the first step in the production of proteins from genes. Mutations in the AFF4 gene are thought to cause accumulation of the AFF4 protein. The excess of the latter interferes with transcription, with consequent problems in the development of various organs and tissues, which give rise to the typical symptoms of CHOPS. The same mechanisms are involved in the cohesins, whose mutations are responsible for other syndromic condition known as "cohesynopathies", of which Cornelia de Lange syndrome is a part.

This syndrome was discovered in 2015 by Dr Krantz and Dr Izumi and it count only 34 cases documented around the world.

This symposium will be an opportunity to explore and debate on the new horizons for CHOPS syndrome, to accelerate research and therapy development.

THIS EVENT IS SPONSORED BY

Children's Hospital of Philadelphia

COOL CARS FOR KIDS

FONDAZIONE CHOPS MALATTIE RARE

CHOPS SYNDROME GLOBAL

CLINICIANS, RESEARCHERS AND PARENTS/CAREGIVERS

PLEASE REGISTER TO JOIN THE SYMPOSIUM

ALSO FOR REMOTE PARTICIPANTS

https://chopssymposium.sciencesconf.org/

Follow these instructions -

There are different links towards the left of the main page/menu.

- Click on the button “Registration”
- Create an account (you will receive an email with an activation link) or login if you have an account with SciencesConf.org or HAL already
- Click on the link and complete your account creation
- Then click again on the button “Registration” on the MAIN MENU of the left column to register

If you are attending the symposium as an expert of the field, please indicate in the registration form which is your institution and if you will join IN PERSON or REMOTELY.

If you are a parent/caregiver, there are some opportunities which the organizing committee of the symposium is offering. Please indicate in the registration form the number of accompanying persons (i.e. 3 if you are coming with your spouse/partner and your two children) and INDICATE IF YOU ARE WILLING TO HAVE MINI CONSULTS WITH EXPERTS. You can indicate more than one option. Once you registered, please go to the section “Mini consults Sign up” that you find on the left column in the main menu of this page.

It is important to know this information in advance in order to properly plan the event.

Friday, July 19, 2024

TIME	EVENT
08:30 - 12:10	CHOPS Syndrome - clinical aspects (Conference Rooms in Abramson 123ABC)
08:30 - 09:00	Welcome and introduction - Lainey Moseley and Manuela Mallamaci
09:00 - 09:25	Genetics - Dr. Ian Krantz, Sarah Raible, MS, CGC
09:25 - 09:50	Gastroenterology - TBC
09:50 - 10:15	Cardiology - Dr. Catherine Avitabile
10:15 - 10:30	Coffee break
10:30 - 10:55	Pulmonology - TBC
10:55 - 11:20	Child Development - Dr. Mary Pipan
11:20 - 11:45	Speech & Alternative Communication methods - TBC
11:45 - 12:10	Immunology and inhibitor treatment therapy - Dr. Alix Seif
12:10 - 12:35	The communicative-linguistic and oro-alimentary function development in subjects with CHOPS syndrome - Dr. Gemma Levantino
12:35 - 14:00	Lunch
14:00 - 17:00	Family consultations with experts (Conference Rooms in Abramson 123ABC)

Saturday, July 20, 2024

TIME	EVENT
09:00 - 12:30	CHOPS Syndrome - Research and perspectives for a cure (Conference Rooms in Abramson 123ABC)
09:00 - 09:15	What it means to be a family advocate for rare disease - Lainey Moseley
09:15 - 09:30	Fondazione CHOPS Journey - Manuela Mallamaci, PhD
09:30 - 09:45	The scientific committee of Fondazione CHOPS: our task and our goals - Dr. Valentina Massa
09:45 - 10:00	Roadmap to clinical trials for CHOPS - Dr Neil Hackett
10:00 - 10:15	Coffee break
10:15 - 10:35	Molecular mechanism of CHOPS syndrome: Insights from novel AFF4 variants (Conference Rooms in Abramson 123ABC) - Dr. Kosuke Izumi
10:35 - 10:55	Deciphering AFF4 expression dynamics: insights into neurodevelopmental disorders and targeted interventions - Dr. Ilaria Parenti
10:55 - 11:15	MeCP2 interacts with the super elongation complex to regulate transcription - Dr. Jun Young Sonn
11:15 - 11:35	Path to a cure in rare diseases and CHOPS - Ethan Perlstein, PhD
11:35 - 11:55	Patients First: Rapid Prototyping of Repurposed Drugs for CHOPS Syndrome - Richard Novak, PhD
11:55 - 12:15	Super elongation complex and study of disruptors/inhibitors of the complex and their use for treatment of cancer and other diseases - Dr. Shilatifard
12:15 - 12:30	Closing Remarks - Lainey and Manuela
12:30 - 14:00	Lunch
14:00 - 16:00	RARE | A Documentary by Lainey Moseley

MINI CONSULTS SIGN UP

This section is intended for caregivers only.

Please first register to the symposium, then come back to this section and complete the intake form.

There will be an opportunity for families to sign-up for individual brief 15-20 minute "mini consults/meetings" with some of the invited specialist if you have any questions you'd like to discuss about your child. These are ***not*** formal clinical visits – merely a chance to ask more general questions to the providers about your child.

If you would like to sign up for a mini consult, a completed medical history questionnaire is required in advance, you can download it by clicking the following

Intake Form.
Please send back the completed intake form to RAIBLES@chop.edu

A list of specialists attending are below. Please register and send back the form by **June 15th** if you would like to meet with any of these specialists and indicate which ones in order of preference in the registration form.

Specialties:

- Genetics
- Gastroenterology
- Cardiology
- Pulmonology
- Child Development
- Immunology

PLEASE RESPOND AND SEND

FORM BACK BY

JUNE 15

GENERAL INFO

This symposium will be an opportunity to explore and debate on the new horizons for CHOPS syndrome, to accelerate research and therapy development thanks to the effort of experts and families.

You can discover more about family groups by exploring the following websites

https://www.chopssyndromeglobal.org/

https://fondazionechopsets.com/en/

Fondazione CHOPS Malattie Rare (meaning CHOPS Foundation Rare Diseases) was established in May 2023 and it has a medical and scientific advisory board lead by Dr. Ian Krantz. Fondazione CHOPS has already funded 4 research projects.

Read more about its collaboration with the company Unravel Bioscience Inc. by clicking here.

More scientific info about CHOPS Syndrome can be found here:

https://fondazionechopsets.com/en/chops-syndrome/what-it-is/

ACCOMODATION

Some rooms are reserved at **Sheraton Philadelphia University City**, that is close to the hospital where the symposium will take place.

A special group rate is available at $179 per night for either King rooms or double Queens.

Please click this link to book your individual reservations, or call 1-888-627-7071 to make your reservations.

Family groups are scheduled to arrive 07/18/2024 making their own reservations. As a reminder, the cutoff date for reservations is June 18, 2024.

Overnight guest parking at the hotel is $35.

For any issues with your booking online, please contact: lwmoseley@comcast.net

The symposium will take place at the Children's Hospital of Philadelphia, 3401 Civic Center Blvd., Abramson building - 1st floor, Philadelphia, PA 19104.

DATES

Program runs July 19th through July 20th, 2024. Participants are expected to arrive on July 18th and depart on July 21th. Participants are expected to attend the whole programme.

HOW TO GET THERE

1. From Philadelphia International Airport (25 minutes): take 76 West towards Philadelphia, exit on 346 to South Street and then left on 34th street.

2. From 34th Street Amtrak train station (6 minutes by car): take 34th street, 7 miles to CHOP.

3. From NYC and points North of Philadelphia (2 hour drive): take the New Jersey Turnpike South toward I-95, take exit 6 to merge on i-276 towards Philadelphia, take 95 South to Philadelphia, 676 west, to 76 East, exit on 346 onto South Street, left on 34th street.

PRIVATE MEDICAL AIDS

In some countries parents are struggling to be assisted by their private medical aid due to the fact that there is no Code for CHOPS Syndrome.

The following information might be of assistance –

2024 ICD-10-CM Diagnosis Code Q87.1 (it came into effect on 1 October 2023.) It is the American ICD-1-CM version and may differ in other countries but gives a basis.

Congenital malformation syndromes predominantly associated with short stature.

Under the code Q87.1 are multiple codes that contain a bigger level of detail.

ICD - acronym for International Classification for Diseases

Q87.1	CONGENITAL MALFORMATION SYNDROMES
Q87.0	Other specified congenital malformation syndromes affecting multiple systems
Q87.11	Prader-Willi syndrome
Q87.19	Other congenital malformation syndromes predominantly associated with short stature
Q87.2	Congenital malformation syndromes predominantly involving limbs
Q87.3	Congenital malformation syndromes involving early overgrowth
Q87.4	Marfan syndrome
Q87.40	….. unspecified
Q87.41	Marfan syndrome with cardiovascular manifestations
Q87.410	Marfan syndrome with aortic dilation
Q87.418	Marfan syndrome with other cardiovascular manifestations
Q87.42	…. with ocular manifestations
Q87.43	…… with skeletal manifestation
Q87.5	Other congenital malformation syndromes with other skeletal changes
Q87.8	Other specified congenital malformations syndromes, not elsewhere classified
Q87.81	Alport syndrome
Q87.82	Arterial tortuosity syndrome
Q87.83	Bardet-Biedl syndrome
Q87.84	Laurence-Moon syndrome
Q87.85	MED13L syndrome
Q87.89	Other specified congenital malformation syndromes, not elsewhere classified

OMIM CHOPS SYNDROME

A number sign (#) is used with this entry because of evidence that CHOPS syndrome is caused by heterozygous mutation in the AFF4 gene (604417) on chromosome 5q31.

WHAT DOES OMIM ENTAILS

OMIM is the acronym for Online Mendelian Inheritance in Man, a continuously updated catalog of human genes and genetic disorders and traits, with particular focus on the molecular relationship between genetic variation and phenotypic expression.

(https://www.omim.org)

RESEARCH ON CHOPS

Fondazione Chops ETS piloted the research with UNRAVEL BIOSCIENCE. They are using artificial intelligence computing to investigate drug compounds that can help the patient.

24 CHOPS and 8 Cornelia de Lange patients enrolled for the research.

SPEECH research was also started, with Dr Gemma Levantino from University of Palermo, Italy heading it, approved by Fondazione Chops ETS.

Two questioners were sent out for parents to complete.

Results will be discussed during the July Philadelphia Symposium.

Jackson Lab is working on mice models concerning CHOPS.

A dedicated geneticist is researching the **mechanisms that cause** CHOPS.

FUNDING FOR RESEARCH

The commission of FONDAZIONE CHOPS ETS, lead by dr Ian Krantz, awarded 3 scientists with grants (all funded by Fondazione chops). The first 200 000 US dollars and two seed grants of 20 000 US dollars each.

For Mario ……. and not only

A huge appreciation for all the fundraising done and the donars

to make this possible.

2024 - CONFERENCE 19 & 20 July

You can still decide to join the symposium in person or remotely.

THE CHILDREN'S HOSPITAL OF PHILADELPHIA, USA.

There will be a few presentations to explain the progress our Scientific Committee has made towards a treatment roadmap.

A few experts that will present new data about CHOPS Syndrome.

Remember to RSVP/ register on the web page

https://chopssymposium.sciencesconf.org/

Lainey Moseley - lwmoseley@comcast.net

Research with Unravel Bioscience – ongoing

Initial results will be made available during the Symposium

ONGOING RESEARCH - in joint venture with

Unravel Bioscience

CONTINUOUS FUNDRAISING NEEDED – 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