CHOPS SYNDROME NEWSLETTER

IN HONOUR OF

AUSTIN ORME 9/12/2022 – 31/7/2023

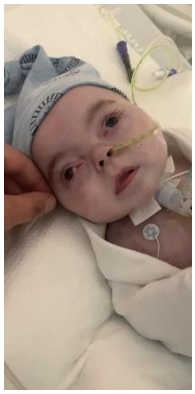
REST IN PEACE LITTLE ANGEL

To the CHOPS community

I am extremely proud that the newsletter this month is honouring my sweet Austin. His passing left a very big hole in my life and in my heart. The CHOPS community has been amazingly loving and supportive throughout this difficult journey, and I cannot express my gratitude enough. Although everyone's sentiments and thoughts do not fill the holes that Austin's absence have left, they help and mean so very much.

Austin passed away from complications relating to chylothorax. This is a condition that developed after his lymphatic system was damaged - likely during his first heart surgery. It caused fluids comprised of fats, proteins, and immunoglobins to drain into his chest cavity. Our doctors tried to fix the damage through multiple surgeries. However, nothing worked. After months of battling, we ran out of options and we had to make the impossible decision to remove his life supports and let him go.

As all CHOPS children are, Austin was very special. He was unbearably cute – his big blue eyes had



a depth to them that revealed so much about the beautiful soul inside. The first thing that most of his care staff and visitors would notice were his long, gorgeous eyelashes. They may have been the only presentation of CHOPS Syndrome that did not create a struggle for him! Austin was very small – he weighed just under three pounds when he was born at 35 weeks, and only nine pounds when he passed away on July 31st at just under eight months old. I also must mention his little hands – he loved hand-holding. The first thing I would do when I went to the hospital everyday was announce "Hi baby!" and find his hands. No matter how sedated he was, I always got a response and usually he would open his eyes to peek at me. Austin used a soother for a short period of time, but abandoned it after he learned this adorable lip suckling technique that he did almost constantly. Often the only sound in his room (other than the machines) was the smacking noise of him sucking his bottom lip.

Austin, like the rest of our little family, loved cuddles and snuggles. Holding him was my favourite thing in the world. I think there were maybe only three or four days out of his 250 days that he wasn't held for hours at least once in the day. It was not easy – primarily because of his tracheotomy and ventilator – but we always made it happen. His last day with us, he was in our arms the entire day and until the very end.

After Austin was diagnosed with CHOPS Syndrome, I spent a lot of time worrying. I worried about having a disabled child and how I would handle it. I worried about the practical and logistical implications of having a child with a tracheostomy. I worried about how I was going to balance my career with being his primary caregiver. I worried about how my other two small children would adapt to having a brother who was often sick and needed a lot of my time and attention. I worried we would never be able to fulfil our dream of one day moving to our remote farm due to distance from a hospital. Most of all, I worried about Austin and the multitude of challenges he would face due to his condition and related health and physical issues.

Now I would give anything to have those worries. Now all I wish is that I had a future with him. But in a way, I feel like I get a piece of this future through your CHOPS kids. There is truly a connection between these special children. I see him in them. I feel Austin continues to live on in his CHOPS brothers and sisters. And although I mourn Austin, I am so grateful for all of the other CHOPS kids and the joy they bring to so many people's lives, including mine.

Love, Angela Orme

CHOPS SYNDROME RESEARCH

Big thanks and gratitude to Manuela Mallamaci (founder) and Lainey Moseley for all the effort they had put in to start up the Foundazione CHOPS Mallatie Rare research program.

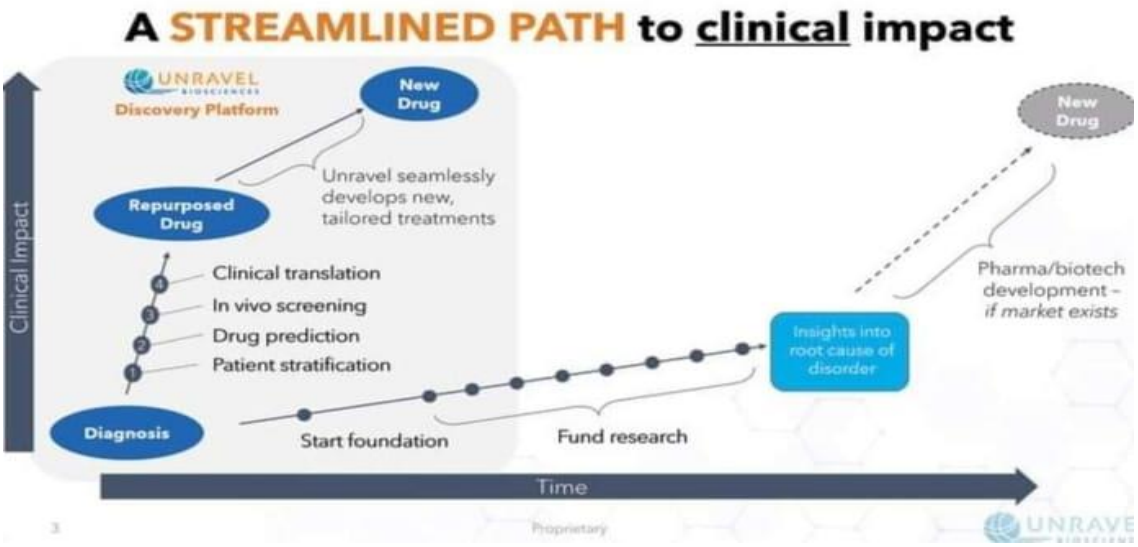
The medical and scientific advisory board members are - Dr Ian Krantz - President, Dr Valentina Massa - Member, Dr Emanuela Scarano - Member, Dr Katsuhiko Shirahige - Member, Dr Eleonora Orlandini - Member, Dr Neil Hackett - External consultant.

(for more information – https://fondazionechopsets.com/en/pages/la-commissione-scientifica)

The research will be carried out by UNRAVEL BIOSCIENCE. Our kids will have the opportunity to be directly involved in the collection of biological samples. Some Cornelia de Lange families will be involved as well.

A virtual information sessions was arranged by Manuella Mallamaci. A few parents (and Luigi) joined. Representatives from Unravel Bioscience attended.

Richard Novak (CEO, Unravel Bio) gave a brief preview of the research planned where RNA nasal swobs from patients and sex-matched parent/sibling will be used.



The main aim initially will be to find repurposed drugs to alleviate various symptoms for the patient. Research will be applicable to help our children.

The meeting and presentations were recorded and will be available for parents who could not be join during the virtual meeting. Please follow …..

https://drive.google.com/drive/folders/12_a24muwY5pqa5P8vekEj_WM-yEdpNTl?usp=sharing

Please contact Manuella Mallamaci as soon as possible if you are interested to have your child to partake in this research study

info@fondazionechopsets.com

Fondazione CHOPS ETS will be ready to fund this project. An amazing effort was launched by Mario's and Luce's family and their teams to raise funds to the amount of 230 000 euro. These funds will contribute partly to enable the first phase of the research to commence.

IMPORTANT

We need all possible assistance to raise funds - please spread the word and try to get as many funders as possible in your own country to contribute to this very important research.

Although our children have an extremely rare syndrome, it is never too rare to care.

WE CARE.

There are now 36 people known who had been diagnosed with CHOPS syndrome.

Awareness can ensure that those struggling with symptoms and not yet been diagnosed, can be drawn into the circle and receive assistance, guidance and comfort from other parents and children who had been on the CHOPS path.

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 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.

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

CHOPS FOUNTATION GLOBAL

ive.rarevillage.org

Research body

To donate, go to

chopssyndromeglobal

Click on DONATE button,

complete detail, specify it is for CHOPS,

choose the monetary value you are donating in.

ITALY (EUROPE)

FONDAZIONE CHOPS ETS

**It is possible to donate via bank transfer to:

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

OTHER COUNTRIES

givealittle.co.zn

IDENTIFY

**ITALY BANKING DETAILS

Gofundme.com

IDENTIFY

**ITALY BANKING DETAILS

BackaBuddy (South Africa)

IDENTIFY

**ITALY BANKING DETAILS

LIAM'S STORY

Hello! My name is Kathleen, I am the mom to an always smiling, ornery, smart, funny, loveable 22-year-old who I call my little miracle and determined boy named Liam.

Throwback Thursday , Liam coming home from NICU Unit 21 YEARS AGO! Then a few months later in PICU for 4 months , my hero. ❤️

Liam was the first boy and only twin in the world diagnosed with a very rare genetic disease called CHOPS syndrome. CHOPS stands for cognitive delays, heart issues, obesity, pulmonary issues, and short stature. LIAM was in a study at Children Hospital of PHILADELPHIA for years ALONG with 2 other girls.

THEN after years one day I got the call in 2015 , "the doctor says we found out what Liam may have". This is the day we been waiting for. NOW There are only 4 kids in the world diagnosed with this condition. Some are as young as a 6 months old. There is no treatment currently, however.

There is cutting-edge research going on giving families hope. This could help the younger kids not to have the damage that was done to Liams small body, height of only 3'9.

When Liam was born he required many hospital stays of over a year combined. Many, many surgeries. He required a ”trach" from 4 months old for 4,5 years, had 2 reconstruction airway surgeries due to very narrow airways. This year he had low platelets (44 000) and thyroid.

This is a glimpse in A-day in the life of Liam and as his mommy and caregiver. His vitals are checked his pulse o2, and hr etc. He requires 2 x's a day nebulizer treatments, along with 5 different medications, paired with chest vest therapy to help stop the mucus build up in his lungs, to help to keep away aggressive lung infections.

Then his day continues with lunch, and on nice days take him out for a walk or on an adventure or sit on back patio. In the evening dinner is made, then repeat all his nebulizer treatments, vest therapy, and his oral medication. At bedtime he gets a bath and has to be changed.

Again this is what rare disease day is all about to bring awareness . When some of these adult kids age out they have no place to go during the day. It's so hard on parents especially me being a single parent. Liam needs one on one on most and all of his daily living needs. I am lucky I have some nursing help to assist me.

He also has chronic Lung disease and a very small airway which required 2 major airway surgery. Along with that, has heart defects which required surgery, he is non-verbal, visually impaired, hearing loss, skeletal issues, short stature reflux and many other issues.

It has not been an easy ride with Liam's medical issue but one thing throughout his medical journey he always smiles and " To see him smile makes our Lives worth-while." Along with school and Liam's prior teachers and his aid Terry Simpson from Child and Career centre in Coatesville, we helped Liams with his skills in sign language. He is able to communicate along with his iPad speech machine.

Liam wants to be able to raise funds for all his CHOPS friends, mainly the younger ones that will help with stopping the progressive of this disease, AND WE NEED FUNDS TO DO THIS. The older our kids get the less chance that any interventions will help.

FDA drugs are being repurposed to help kids with rare diseases. But the research to discover these drugs for CHOPS is expensive.

Our community is trying to raise 300K so that we can partner with a research organisation. If we can find a drug that can slow down the progression of CHOPS Syndrome, there is potential to improve our kids' verbal skills, lung disease, growth and intellectual abilities. But the clock is ticking for THESE PRECIOUS CHILDREN. Will you please consider a donation to help Liam and all the other families in our CHOPS community in this fight to save our kids!

If you could donate any amount, we would be so grateful. To date, our community has raised over $75k but we need your support to get over the finish line. Small amounts matter as well. Lots of people ask “What can I do for Liam’’, well now is your time to donate directly into banking options as shown in above page.

For more information on CHOPS, visit our website:
https://www.chopssyndromeglobal.org/

https://fondazionechopsets.com/en/pages/la-commissione-scientifica)

FRANCISCO'S PATH WITH CHOPS

Francisco was born at 34 weeks with blood and water in his lungs. He is now 3 years old.

He cannot crawl or walk yet but he is receiving PT. He drags himself forward in stead of crawling and rolls everywhere. He is using a walker.

As a baby he was constantly struggling with a lot of reflux and chocking. He was given Nexum, apparently not helping him. He was also on Estoriodes

(prednisolone which the pulmonologist increased).

Fortunately Francisco has no heart defect but has difficulty with the following - Anemia of prematurity, BPD (bronchopulmonary displaysia), Grade 2 IVH of newborn - resolving, Underactive thyroid, Patent ductus arteriosus, Prematurity 2000 – 2499 grams born 33 – 34 weeks, MDRO Multiple drug resistant organism, genetic disorder, Bronchiolitis.

He loves to play with his siblings. He gets sick very easily perhaps with siblings bringing viruses from school which ended him up in ICU with rhinovirus and needed extra support. It is difficult to keep him healthy and without ailments.

CHOPS AND SPEECH ABILITY

So few of our kids have the ability to speak.

Some of them have a small vocabulary of a few word but cannot have a conversation.

It has been noticed that individual kids will use a word like "Mamma" a couple of times and then not ever use it again. Thus, they have the ability to speak but looses the words somewhere along the line.

Quite a few parents have already taught their kids sign language.

How good it will be if all newcomers to sign language can learn the same type of sign language which will give our kids the opportunity to communicate with one another virtually or when they meet.

Unfortunately there are so many types of sign languages used in different countries.

There is however MAKATON, a sign language rooted in United Kingdom and used across many countries. MAKATON is used to help **hearing people** with learning or communication difficulties. BSL, ASL and other sign languages are where people are hard of hearing or deaf.

Then I noticed that some of the kids have an iPad speech machine to assist with communication. How effective is this method?

Some kids are using flash cards to communicate.

It will be great if all parents will send me a message to issteyn@yahoo.com in order to advise newcomers to CHOPS what type(s) of assistance was given to their kids and what works best.

Your advice and input will be valuable to assist our children and families.

EIDER'S LIFE …… A PATH FULL OF CHALLENGES ……. THE SKY IS THE LIMIT!

Life has never been easy for Eider, even before birth. Results in the amniocentesis were normal but doctors already saw at 3rd echography that measures did not fit in with the gestation time. Since then, pregnancy was kept under close observation and **I** was hospitalized until Eider was born, 15th November 2006. She was such a beautiful girl, with big opened eyes and long eye-lashes… Apparently, everything went alright**.**

However, time went by and Eider seemed not to have a normal development. Furthermore, Eider was also diagnosed with hypothyroidism when she was six months old and then she started a treatment with L-thyroxin. She spent her first year surrounded by doctors, nurses, injections…she was always ill with breathing problems. She was not strong enough to eat by herself, so she did not put on any weight. Whenever we went to see her pediatrician, he repeated that "*We had to accept that she would never be able to walk, speak … properly. It was time to assume Eider will be like a vegetable. Life was cruel but we simply had to accept the situation*".

When Eider was about to reach her first year, she was really ill. We went to the emergency department of another hospital. Eider was once again hospitalized immediately. Doctors found out she had a congenital heart disease that had not been detected before: her *ductus* was still open and she had several VSDs (Ventricular Septal Defects). She was critically ill and she underwent an urgent surgery intervention. We spent 3 long months in hospital . They closed the *ductus* and put a banding around pulmonary artery to relieve the consequences of the VSDs which could not be closed as they were impossible to reach. As our daughter was not strong enough to eat by her own, enteral feeding was the only solution. We used a nasal-gastric tube connected to a machine to pump the milk. Thanks to that, she slowly started putting on weight. Eider was fed like that for six months.

At the age of four, she started a growth hormone treatment to solve her growing problems. At birth she measured only 43cm. She was always shorter than children of her age. In the meantime, doctors began to pose questions about her case: Eider's phenotype was really particular, with some features which reminded Cornelia de Lange syndrome with severe physical and psychic delays. A large number of genetic studies were done but all of them showed negative results. Nobody knew what caused her symptoms. So, from a medical point of view, Eider was still a big mystery and her life-quality expectancy was really low. But we always believed in Eider's ability to learn. That is why we never stopped stimulating her. We made a continuous effort in order to improve our daughter's abilities. We tested all kind of therapies: early stimulation, physiotherapy, osteopathy, speech therapy….and so on.

Thanks to an article published in People's magazine, a friend of us saw Leta's picture on Facebook. Eider and Leta looked so alike that we were shocked when we saw her. We quickly got in touch with Dr Krantz who accepted to review Eider's medical records and to enroll our daughter into a research program to test for CHOPS syndrome. We used to travel to Necker Hospital for children in Paris every year. So we coordinated shipment of some of Eider's DNA already stored at Necker to be sent to Children's Hospital of Philadelphia. At that time, there were only three other cases of Chops. The 23rd July 2015, we received an email saying that Eider had a *de novo* change in AFF4 gene, confirming the diagnosis of CHOPS syndrome. She was the 4th case in the world and the 1st case in Europe. We could at last name Eider's disease and we were now sure that we had not been the ones who transmitted the disease to our daughter. Actually, we had been feeling so guilty……

In 2018 Eider underwent another surgery intervention. The banding which doctors put around her pulmonary artery in 2008 restricted blood flow too much, it was time to remove it. Eider had grown during those 10 years and it was absolutely necessary to do it before serious problems arose. Apparently it was an easy intervention and we were supposed to be at home 15 days later. But everything that could go wrong did. After a 9 hours surgery Eider was again at the thin border between life and death. She spent 3 months in the Intensive Care Unit, and after a tracheotomy she began to recover slowly. Finally she returned home with oxygen therapy because she was still not able to breath on her own. She had to learn again to sit, to stand up, to walk, to speak … everything from zero. Some months later her lungs worked better and she could breathe on her own. Our Eider had won the battle again. Our children have fragile bodies but a strong personality and a big desire to live.

Contrary to what doctors had told us, Eider's every day's life has changed a lot. Eider is able to walk, run and jump although with some difficulties. She speaks and understands Basque language and Spanish. She has spelling problems and we try to assist her with language therapy. She has always been to a "normal" school. We have always prioritized Eider going to normal school in an inclusive way. While she was in Primary School, she was in class with other children but had a person to support her fulltime and had an adapted curriculum. Her classmates have always been her learning engine, an example to look at. She's been loved by everyone. She's been a very lucky girl. Eider now attends a normal High School where there is a classroom named "Stable Classroom" where she goes with other students with Special Educational Needs. Eider is such a happy girl, full of live. She loves dancing, singing, swimming and being with her schoolmates. That is why we have always taken her to different kind of activities along with other children such as musical and artistic Summer and Easter camps. Nowadays, she attends Basque dance courses, she goes to paddle surf/ski, learns swimming and receives therapy with horses. It is our priority that Eider takes part in various activities in an inclusive way with other "normal" kids who stimulate her constantly. We always manage to engage a person who attends to her in all those activities.

Eider is a happy and loving girl, with a big personality. Despite all the obstacles that life has put on her way, she manages to make us smile every day and she never ceases to amaze us with her daily improvements. She is such a brave girl! She never gives up on the new challenges that life brings. On the contrary, she always faces them with courage and a smile.

This month Eider will be 17, but she still continues to surprise us with words or gestures she learns, or the way she gets excited when we decorate our home for Christmas or Halloween. All these amazing activities help to overcome the hard times.

Our message can only be positive. No one can predict what the limit of our children's abilities will be, how far they will get. The limit is the sky! As parents, we can only try to reach the highest, to reach for the sky, and believe we can make it, working every day and with a lot of imagination and patience. All this effort is worthwhile when we see the **pride** in our daughter's little face for achieving another milepost. **LITTLE BY LITTLE EIDER IS REACHING FOR THE SKY**!

AIMS FOR THE NEXT TWO YEARS

2024 - CONFERENCE 19 & 20 July

Keep the dates open for the Conference in Philadelphia which will be held at

THE CHILDREN'S HOSPITAL OF PHILADELPHIA.

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

It is expected to have a few experts that will present new data about

CHOPS Syndrome.

Planning for a fun outing to the Philadelphia Aquarium or Franklin Museum.

Remember to RSVP

as soon as possible, latest end January 2024

Lainey Moseley - lwmoseley@comcast.net

At least 12 families should attend to ensure continuation.

EXPAND RESEARCH - in joint venture with

Unravel Bioscience

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