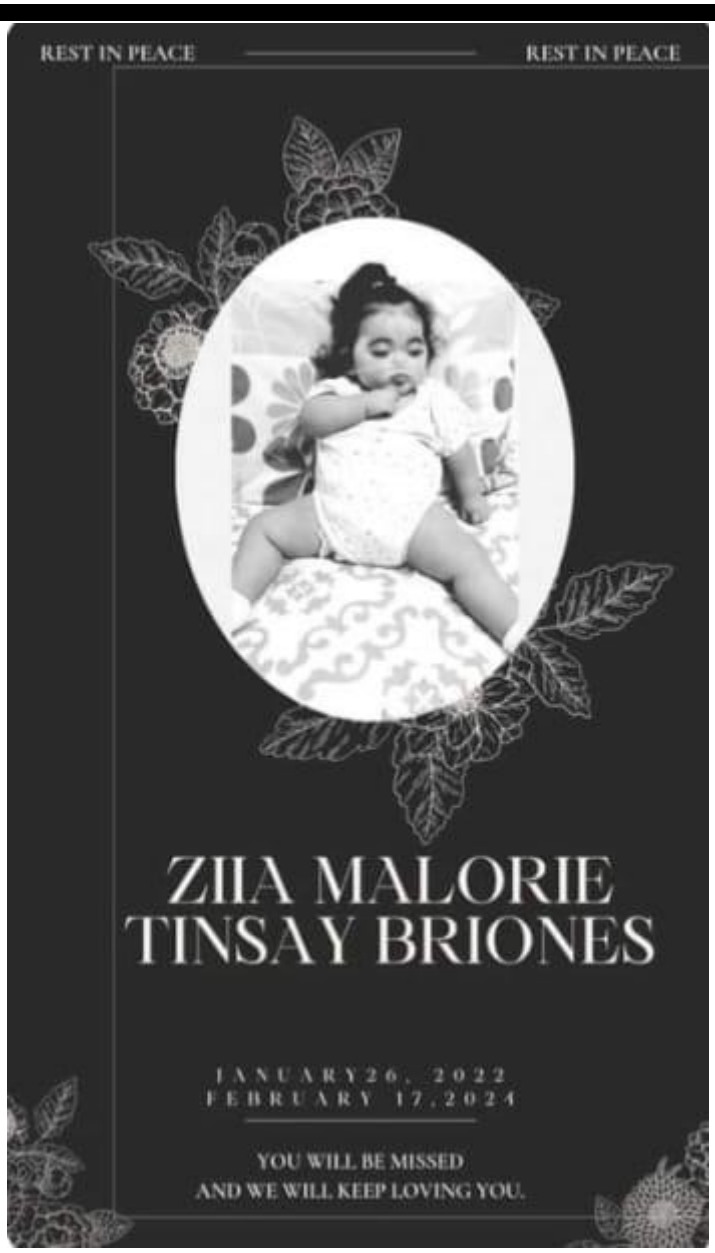
IN HONOUR OF



Our hearts go out to the Briones family for the loss of their darling Ziia.

Ziia fell ill before her second birthday and was in hospital since.

REST IN PEACE LITTLE ANGEL

THE YEAR OF RESEARCH ON CHOPS SYNDROME

2024

UNRAVEL BIOSCIENCES

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.

FOR MARIO

and

all others diagnosed with CHOPS and still to be diagnosed for years to come.

CONGRATULATIONS Manuela

Mario and Luce's parents decided that they need to do fundraising to enable research on CHOPS. Their families formed a strong bond to make the fundraising a tremendous success.

Manuela founded FONDAZIONE CHOPS MALLTIE RARE ETS and the massive effort started to collect funds to make the research possible. Looking for a laboratory to set off the research began and eventually UNRAVEL BIOSCIENCE in US was elected. The initial amount was paid over to UNRAVEL BIOSCIENCE for the first step along the path - the search for repurposed drugs to make the life for CHOPS patients easier and more bearable. FONDAZIONE CHOPS ETS is paying for all the participants to this study.

Thank you ITALY for opening your hearts and purses to enable this research.

THE CHOPS SYNDROME COMMUNITY AND PARENTS SALUTE YOU.

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	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: 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	givealittle.co.zn IDENTIFY **ITALY BANKING DETAILS Gofundme.com IDENTIFY **ITALY BANKING DETAILS BackaBuddy (South Africa) IDENTIFY **ITALY BANKING DETAILS
Click on DONATE button, complete detail, specify it is for CHOPS, choose the monetary value you are donating in.		

OTTILIA

Ottilia was born 8 years ago in February 2016.

When Ottilia was 4 months old, she started to hang around and she couldn't cry because she was so hoarse. When we were sent home again from the hospital, we applied to another hospital where they were absolutely fantastic. It turned out that Ottilia had fluid in her lungs. This was June 2016.

Then began a period with an awful lot of tests, hospital visits, etc. We were hospitalized several times for pneumonia. They concluded that Ottilia had reflux, but they also checked a lot of other things.

When Ottilia was 2 years old, she started to make a lot of rattling noise in her throat. We sought treatment but they said she just had a cold, but she didn't have a cold. Then she started throwing up a lot and drooling more and more...we sought care 3 - 4 times but without success.

In May 2018, we received the chops diagnosis.

In May 2019, Ottilia finally had stomach surgery for her reflux and she has been doing great since then.

Swedish version

Ottilia föddes 8 v för tidings, a fob 2016. Nor Otelia va 2 v började hon rossla/låta mycket för sin hals. Vi sökte vård men dom sa att hon bara va förkyld, men hon va inte förkyld. Sen började hon kräkas jättemycket och rosslade mer och mer..vi sökte vård 3 - 4 ggr men dom gjorde ingenting.

När ottilia va 4 månader så började hon bli hängig och hon kunde inte skrika för hon va så hes så När vi återigen blev hemskickade från sjukhuset sökte vi till ett annat sjukhus där dom var helt fantastiska. Det visade sig att ottilia hade vätska i lungorna . Detta va juni 2016. Sen började en period med oerhört mycket tester, sjukhusbesök osv. Vi låg inne på sjukhus flertalet gånger för lunginflammation. Dom kom fram till att ottilia hade reflux men dom kollade massa andra saker också. I maj 2018 fick vi chopsdiagnosen.

I maj 2019 fick ottilia äntligen operera magen för sin reflux och sen har hon mått jättebra:)

RESEARCH EXCITEMENT

The parents of 24 kids/adults with CHOPS had signed up to partake in the first part of the Unravel research. A few parents of kids with Cornelia de Lange syndrome are also signed up for the research.

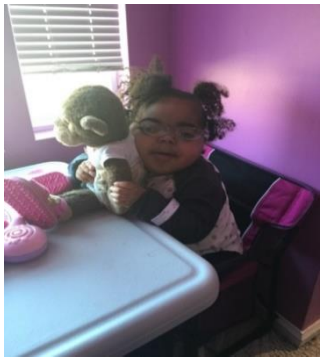
Those who interacted with Unravel, received nasal swabs which was sent via FedEx and the samples were sent back in the same manner. At present a few are still outstanding.

We wish for a relief to all those living with CHOPS and Cornelia de Lange syndrome.

Luigi, Lainey and Leta, Luce, Eider, Salvador, Teddy, Gianni and Mario

TAYLOR

An Article in People Magazine Changed Our Lives



IUGR, 2 vessel cord, and polyhydramnios are just a couple of the words that were consistent throughout my pregnancy. I was hounded by genetics and endured monthly sonograms all while trying to enjoy my pregnancy. After spending 8 months in the NICU, Taylor came home in August of 2008 with a tracheostomy for subglottic stenosis and without a formal diagnosis but clearly an underlying genetic condition.

Years later while sitting in her room I saw a picture of a little girl who looked just like Taylor in an article in People Magazine. Interest peaked, I began to read the article and noticed the similarities between the children in the article and Taylor. I was even more shocked to learn that the research was taking place at CHOP as that is where Taylor spent the majority of her time in the NICU. In 2015 I emailed Dr. Krantz and said, hey, I think that my daughter has CHOPS Syndrome. We are coming into CHOP in a couple of months and I wanted to know if he would see her. He agreed and the moment that he walked in the door, he said absolutely. After many years of unanswered questions and countless tests, an article in People Magazine answered a question that many doctors tried to solve. She was the 8th child in the world to be diagnosed with CHOPS Syndrome.

Taylor is now a 16-year-old diva in her own right. We have encountered many things on the journey to 16 and she has taken it all in stride, including her diagnosis of interstitial lung disease. Although she has endured many hospital stays, procedures, and countless illnesses, her personality shines through in any room that she is in and she does not have a problem telling anyone what to do and how she wants it done. In her mind, everyone works for her and we are all here to please her. She does not let her “inabilities” limit her in any way. In many ways, she is a typical teenage girl that loves to eat, hang out, and listen to Taylor Swift and Meghan Trainor. Her favourite thing to do is kicking her mom out of her room daily. It is our goal to keep life as normal as possible as we travel this journey of life with her. While sometimes exhausting, we take each day in stride. Through all of the medications, appointments, ER visits, and all other things included we continue to find the joy in this journey and celebrate her strength and determination each day.

MILAN

INTERNATIONAL RARE DESEASE DAY

The fundraising route that Gianni and Manuela take for

MARIO

and

CHOPS research fundraising

stretches from Palermo to Reggio Calabria to Caserta to Pistoia and Siena, to Bologna, to Padova and to Milan.

The Rare Disease Day is a global initiative to raise awareness and generate support for everyone who is affected.

This year it will be on 29 February.

If we all make an effort to contribute to Fondazione CHOPS ETS, we contribute to further research.

CAITLIN

In late 2011, we welcomed our second baby girl. She was small and a little bit extra floppy, but the doctors weren't concerned. Despite being born at 38 weeks, she was just under 5 lbs when we left the hospital and wore preemie clothes and diapers for several weeks. She remained in the lower end of the growth charts for many years.

We were blessed and lucky - Caitlin developed fairly normally, albeit with a slightly inturned left leg the doctors promised she'd outgrow (and we now find needs to be addressed more seriously). But as time went on, things didn't seem quite the same with her. She struggled with things like reading, telling time, numbers, and writing, but then would philosophize about the origins of life, God, and humanity even at three or four years old. She was clearly a brilliant, yet struggling little human.

At two years old, she developed a rash across her body. We first thought that it was chicken pox, but after several weeks of increased pain and sickness, they determined it was psoriasis - some of the worst they'd ever seen on a child that young. They put her on a steroid regimen to clear up her skin and after several months, we saw some progress.

When she was five, she began to show signs of lethargy and weakness. She was constantly unwell. Because of a family history, we knew to have her checked for adrenal insufficiency. She was diagnosed with this cortisol deficiency and began to gain weight rapidly. Treatment helped some, but not well enough, and we sought more answers. She has hypermobility and also low muscle tone in her legs and core, which makes it difficult for her to get much exercise, even though she loves to play soccer!

In 2023, Caitlin's endocrinologist recommended participating in a genetic study focusing on the causes of childhood obesity. Considering that obesity runs on both sides of the family, we agreed. What came back was a mutation of the AFF4 gene - the one related to CHOPS Syndrome.

While we've learned that the mutation Caitlin has is on a different section of the gene than most kids who are diagnosed with CHOPS, we've found some similarities in her symptoms and physical appearance. Though she doesn't have the classic CHOPS appearance - her eyebrows are fairly thin and her features aren't as well-defined - she has a thick head of hair, square-ish feet, and short, triangular fingers. She's so incredibly beautiful!

Caitlin hasn't been formally diagnosed with CHOPS because it is nearly impossible to get into a genetics specialist in our area. We are on a waiting list in two different locations, hoping that we might be able to get some more clear answers. In the meantime, she's been diagnosed with a learning disorder related to math computation and dyslexia, and a pragmatic language disorder.

Caitlin loves art, especially anime, and is an incredibly talented and creative person. She has trained our youngest dog to do tricks and loves animals. She struggles with making friends but when she chooses a friend, she's loyal and unconditionally loving. We know that CHOPS or a CHOPS-like syndrome are things that she will need to be aware of in her continuing health journey, but it isn't something we want to hold her back.

We hope to attend the convention in Philadelphia and meet more families who are on this journey, too. The knowledge and support already are incredible and we are so grateful for the folks who are doing the work to help keep our kids safe and healthy. Sarah Reilley

MAE CONLON

Mae was born at full-term and we went straight home from the hospital. Within the first two weeks of her life we were back in hospital a number of times as Mae had failure to thrive and had very noisy breathing, especially when eating. The doctors didn't seem to know what was going on, some of them were not very helpful at all. After a bit of a fight, we were referred to AlderHey Children's Hospital where we found that Mae had laryngomalacia and a stridor. For the first few years we spent a lot of time being admitted to hospital for episodes of stopping breathing or bronchiolitis. Teething seemed to make everything worse. In nursey Mae needed 1:1 support for cognition and learning and this continued into primary school.

A few months ago, we managed to get Mae moved to a school with specialist provision, it was clear Mae wasn't coping in mainstream anymore and she was finding it difficult and constantly compared herself to her peers and their ability, this was having a huge impact on her. Mae had physical, occupational and speech therapy, which she still has now. Mae has a very curved spine, hypermobile joints and a speech and language disorder. Until recently Mae was diagnosed as having a global learning delay as the doctors and specialist had ruled out Autism and later Prada Willi (Mae is always hungry and struggles with her weight). One of our doctors was convinced Mae's condition and medical history was linked and there was something genetic causing it, thankfully she continued to keep looking for the answer. Mae has been under the pediatrician for pretty much her whole life monitoring her chest, which has settled quite a bit, her height (Mae has always been short, but so am I), her weight and other medical issues as they occur. Most recently, Mae has been having investigations for her adrenal gland function, Mae started puberty at a young age.

Mae has had on-going hearing issues and has had grommets and her adenoids removed. At her most recent operation last July Mae's lungs collapsed during her surgery. The doctors now believe with is linked to her CHOPS syndrome and possibly sleep apnea. Our journey with CHOPS has just begun and each time we see one of Mae's specialist doctors we find that we are referred to another specialist. I am fortunate enough to work in Mae's new school so my boss is very flexible with Mae's appointments (which there are a lot). For now, we need to rule out any problems with Mae's heart, check her kidneys and continue to investigate the strong possibility of sleep apnea. It already feels like I am teaching Mae's doctors and I am sure you all know how scary that feels. As a family we are grateful to have found all of you, to have been included in the research with Unravel and to have been able to speak with Dr Krantz. I am sure we will have lots more questions to come but we are glad to not feel so alone know and be part of the CHOPS Kids family.

We always knew Mae was unique but we just didn't realize how unique.

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, USA.

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 29 February 2024

Lainey Moseley - lwmoseley@comcast.net

More information will be sent soon.

Research with Unravel Bioscience

Started off well in 2024 with 23 families participating.

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