CHOPS SYNDROME NEWSLETTER

Please donate to fund research to find treatments

that can help improve the quality of life for our children.

CHOPS Syndrome is an ultra-rare genetic disorder with only 33 known cases in the world. It stands for cognitive delays, heart issues, obesity, pulmonary issues and short stature.

ABOUT THE CAMPAIGN

Mission is to raise money to accelerate research that will find ways to improve the quality of life for our children and perhaps one day a cure. Currently, there is cutting-edge research going on giving families in the Rare Disease communities hope.

Companies like Rarebase, a biotech company in San Francisco, are developing advanced AI technology to discover and develop treatments for the millions of kids worldwide living with a rare disease. The exciting part of this research is there are many already approved FDA drugs 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 company who can help us 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 our kids. The older our kids get the less chance that any interventions will help.

So we need your help with donations!

100% of your donations are tax deductible and will go directly to research. Thank you so much for your support in this fight to save our kids.

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

Lainey Mosely and Manuela Mallemaci are looking to partner that can help find

a FDA repurposed Drug that might have a secondary benefit for CHOPS.

For more information and research, refer to the

April 2019 newsletter of

CHOPS SYNDROME GLOBAL
and
drs Ian Krantz and Kosuke Izumi (discoverers of CHOPS Syndrome;
Sarah Raible, Genetic counselor

FONDAZIONE CHOPS IN ITALY

Manuela Mallamaci , Valentina Biondi and Reina Raffo worked on this logo for fundraising in Italy. Manuella writes:

Mario,

I can tell you what I've seen in this picture from the first moment; I've seen not just Mario. I've seen all those kids with a rare genetic condition who can't express themselves as they really want to. They are like suffocated to wrong entanglement. But under that envelope they are, that's how they are!

We may not always be so sensitive to their cries for help ….

What I see it when I look Marion in the eyes, is like a chalk of energy, a spark that wants to evaporate and it can't. And this is exactly what drives me to do what I do.

Looking at this stylized omino that reminds me of Pinocchio a bit, I saw an uphill path made even more difficult by an unquantifiable weight, a burden symbolized by the invisible content of that green-blue backpack.

I have seen what for me is the meaning of this picture and its beauty for the universal message it contains: the uniqueness of every single child, the duty of us adults to never judge, so as not to end up in a comparison that is not supposed to exist. Because no one knows how many stairs there will be to climb, no one knows how heavy the backpack will be that every child has to carry during their personal growth journey.

Fondazione CHOPS ETS

OUR CHOPS FAMILY HAD GROWN Since 2019

Adrienne from
(Suzy J Candela and Laura Jininex)
7 diagnosed at 3 years

Austin from California
(Angela Orme)
8 months, diagnosed at 5 weeks

Francisco Franco from Texas, US
(Victor and Grizelda)
3 diagnosed at 7 months

Kyrie from California
(Kayla and Dwayne Caceres)
5 at 2 years

Luce from Tuscany, Italy
(Reina Raffo)
16 diagnosed at 10

Lucian from Auckland, New Zealand
(Anne Belle Agulan) 5

Lina from France
(Nana Yaya Linoush Taico, Emily) 5

Luigi from Italy
((AnnaMaria Mucci)

Mario from Reggio, Italy
(Gianni and Manuela)
2 diagnosed at 22 months

Mark from Phillipines
(Elton Ang)

Salvador from Cape Town, South Africa
(Rae-Dawn and Elizly)
2 diagnosed at 14 months

Zarli from Queensland, Australia
(Ryan & Rinny)
7 diagnosed at 4 years

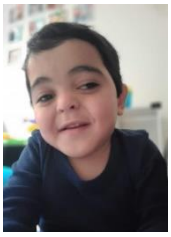
Ziia from UAE
(Edna Tinsay-Briones
1½ diagnosed at 10 months

PHOTO STATION
Names, left to right and top to bottom
Kyrie, Luce, Austin, Fransisco, Lina, Siia,
Adrienne, Lucian, Zarli





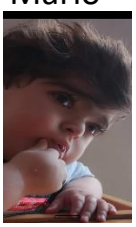




The boys born in 2021

Salvador

Mario



Lucian Carlos Agulan

Hi! I am Lucian, my mum calls me Luy-loi. I am 5 years old and was born full-term at 38 weeks in Starship Hospital in Auckland, New Zealand.

Here is my story: When I was in my mum's tummy at 36 weeks, they found that I have short limbs and a CHD called Coarctation of Aorta. Everyone is expecting that I might not survive but we came through because I am brave! No wonder they call me #BraveLucian; I even graduated NICU in <3 weeks but I have been in Oxygen support since then until today and we still don't know what is causing it.

When I was born the COA in my heart was no longer there but instead, I have PDA (Patent Ductus Arteriosus) which I have gone catheterisation surgery to fill the hole and close it. I also have a Single Coronary Artery; yes, I only have one. Which makes it harder for my Coronary Artery to transport oxygenated blood to my heart's muscles. I am very vulnerable to chest infections; a common cold can admit me to hospital in just a few hours sometimes I get to stay in PICU too. I have had bronchitis, pneumonia, respiratory tract infection and pulmonary hypertension this is why I rarely go out in public specially during colds season. All these respiratory problems are also affecting my heart condition, this is why my mum would always take me to hospital right away.

I love to eat and most of the time I can't stop eating. This is why my mum always makes sure I only eat enough because I also suffer from GERD (Gastroesophageal Reflux Disease) its so bad that it can cause aspirations and leads to chest infection too. I am way too short for my age, which makes me cute really. I cannot walk unassisted; both my feet are over-pronated this is why I need to wear orthotic shoes. I need my walking frame to help me move around but my favourite is to move around with my ride on car because I can go faster. I can only stand if my back is resting on a wall or holding on something sturdy. I can sit with no problem, and I can also crawl. I am not toilet trained yet but were getting there.

I have hearing loss with both my ears, but I can hear you when I'm wearing my hearing aids, which I hate wearing by the way. I am almost non-verbal, but I can communicate with NZSL, picture and word cards; Yes, I can read! My mum taught me when I was just <2yo. I am very sociable and almost always happy; I love having people around me! This is why I am sad that I must stay away from people sometimes to avoid getting sick. I like being silly, dancing, making noise, listening to music, and singing. Playing is my way of learning, my most favourite is sensory play.

Thank you for reading!

CHOPS Syndrome

C.H.O.P.S. stands for **C**ognitive Impairment, Coarse Feature, **H**eart Defects, **O**besity, **P**ulmonary Problems, Short Stature and **S**keletal Abnormality. It is the given name of this severely rare genetic disorder which is caused by a mutation in the genetic material of the AFF4 gene inherited in autosomal dominant manner.
(Izumi, K, Krantz, I. D. et al, 2015)

To date there are only 28 patients diagnosed by genome sequencing. To get to know more please visit our website and pages:
www.chopssyndromeglobal.org

Facebook.com/CHOPSsyndrome

Please **donate** to give-a-little to help us fund Scientific Research to discover and develop treatments for me and other CHOPS children:

(SCAN QR CODE BELOW)

https://givealittle.co.nz/cause/lucian-and-chops-syndrome

@BraveLucian

Laney Moseley is working on the development of a documentary on rare disease and recently partnered with CombinedBrain.

'RARE' unveils what stands between the families fighting rare disease, hoping for treatments, and the emerging groundbreaking therapies available.

The documentary trailer can be seen on www.tooraretocare.com

Please spread the word in order to attract more doners funding research on rare disease.

For more information, contact araredocumentary@gmail.com

The Filmmakers

Lainey Moseley
Producer

Lainey Moseley is an Emmy® award winning journalist who has worked for CBS and NBC News for the past 30 years. She is also the founder of CHOPS Syndrome Global; an organization raising awareness for her daughter Leta's rare disease CHOPS Syndrome.

John Beder
Director

John Beder is an Emmy® nominated and award winning documentary director and producer. John's latest project explored pediatric palliative care and was part of the the Oscar-winning New York Times Op-Doc series.

We need each family to become campaigners for fundraising.

KYRIE

“Hello! My name is Kayla and I am the mom to Kyrie - a 5 year old boy with CHOPS syndrome. Kyrie’s dad ‘s name is Dwayne and we all live in Southern California.

Kyrie was diagnosed with CHOPs at 2 years old. Like most of the families in our community, before his diagnosis he was a big mystery. During my pregnancy Kyrie’s long bones were not growing at a normal rate so our doctors knew something was up. At first, one doctor told me he wasn’t going to live because his chest size was so small!!! Of course that was the worst news I’ve ever received in my life. But then we met with another one that said he would live but would most likely have dwarfism. At that point I didn’t care what kyrie had, I was just so grateful he was going to live.

Fast forward to his birth, when he was born he looked “normal.” His arms, legs, and body were proportionate so the doctors thought “maybe he doesn’t have anything.” However, the first challenge we found was that he had bi-lateral hearing loss. Then at a month old he needed a heart surgery. And after surgery he had some complications and needed oxygen support until he was 10 months. I knew something was up but there was still no diagnosis.

Our geneticist thought he might have kabuki syndrome or Cornelia de Lange syndrome but when we tested for those, it would come back negative. Kyrie was stumping everyone. It wasn’t until he was almost 2, we decided to test all of his genes and that’s how we found a mutation on the AFF4 gene and got lead to CHOPS!

After joining the Facebook group and getting connected to the other families, we didn’t feel so alone. It’s like we have brothers and sisters all across the world. It’s fun seeing everyone’s updates and cheering each other on for all the great progress our kids make.

Kyrie is fun-loving, mischievous, independent, curious, caring, and very determined. He’s been in some type of therapy services since he was 2 months old. Now he’s doing speech, physical therapy and occupational therapy.

It’s been a long road but he’s made a lot of progress. This September he will be starting kindergarten. He loves school and to learn. He’s not so sure on how to play with peers but he LOVES to play with older kids and adults. Just like all the kids I see in our group, Kyrie lights up any room he walks into. Our kids are beams of joy and really teach us how to live!

Thanks for reading our story and can’t wait to get to know you!!“

Best, Kayla

FUNDRAISING CHAMPIONS

The Mallamaci and Raffo families are boots and all into Fundraising for research on CHOPS and ways to make It better for the persons living with this syndrome and support to affected families.

More than €300 000 was accumulated by launching a variety of activities, projects and public awareness via public media.

The challenge is there for other countries to raise to raise this amount to enable more strengthen the power of research towards medication and a cure for this syndrome.

Contact Manuela Mallamaci

Contact Manuela Mallamaci
for more information concerning
fundraising and tax deductions for funders.

Reina and Luce

Mario with Mom and Dad

Thanks to Kathleen Johnston and Kayla Caceres who started individual GO FUND ME fundraisers.

More detail on these and other fundraisers will be given in next newsletter.

Every bit helps!

ZARLI

Hi my name is Zarli, I am a delightful 7 year old who has CHOPS Syndrome. I live in Queensland Australia where I am the only child in my state diagnosed. I go to a wonderful Special School and have various therapies weekly and some special intensive therapy blocks.

The start to my life was a bit of a rollercoaster, respiratory illnesses, hearing loss, cataract surgery, Adenoid surgery, Grommet (ear drum tubes) surgery, feeding issues, growth issues, low muscle tone and Global Development Delay. It all made sense to my family when I received my diagnosis of CHOPS Syndrome just before I turned 4 years old.

I have had some scary times when lung infections land me in hospital and my most recent experience of a tension pneumothorax, where my left lung had completely collapsed due to undetected pneumonia. I had 3 weeks in hospital as I also developed pleural effusion (pus pockets) around my left lung. As scary as this was, I had come out stronger and cheekier than ever.

I have my family support me in my everyday life, my Mum, Dad, big brother and little sister. I also have 2 amazing friends who come and help care for me in my home and take me out on adventures. I am learning to play with my siblings in various ways, and am showing them love by giving them whole body cuddles. My little sister loves to swing and spin with me, one of my most favourite things to do. No matter how fast I spin, I never get dizzy. I also love toys with lights, especially ones that move and sing. I love to play with them on repeat, so everyone in my family sings along with my toys. My favourite things with my brother is when he helps with my nappy (he holds my hands and sings to me) and sitting next to him in the car I love to play and cuddle him.

I am non-verbal and cognitively impaired, however I make the most amazing sounds. I do say Mum, I am starting to say Dad, and I like to make a sound ‘mim’ when I am unhappy. I am starting to communicate in other ways too, I can sign ‘more’ in my own way, I use pictures/photographs of things where I either tap the picture or I take it off my board and hand it to my family to indicate what I want to do. My favourite photos to use are what food I want to eat, when my nappy needs a change, my Mum’s car as I love car rides and of course the photo of my swing. I have hearing loss, I am meant to wear hearing aids but I really don’t like them, as much as my Mum & Dad would love me to wear them, I would prefer to chew them. I also have to wear contact lenses, I don’t quite like having them removed (apparently they need to come out to be cleaned) but I do so love when they are being put back in. Sometimes I even drag my special mat to the table to tell Mum & Dad I am ready for them to go in and I giggle with joy. With them I can see so much!

I get to do some amazing therapies and I show excitement when I arrive at their places. Sometimes I cooperate and other days I like to challenge them as they challenge me. They have helped me in many ways though, and hopefully one day soon I will be able to walk on my own. That will really change what mischief I can get up to!

ZIIA

My name is Ziia Malorie, a.k.a. Zii. I was born on January 26, 2022, at Medeor Hospital, Abu Dhabi, United Arab Emirates.

I was 33 weeks and 5 days inside my mom's tummy when the doctor found out that I was no longer growing and that the oxygen supply to the placenta was weak. That's why they decided to do emergency CS at 34 weeks. The doctor said I was in critical condition and needed to be put on a tube so I could breathe properly because my lungs were collapsed. I stayed in the NICU for 2 months, and a month later, I was re-admitted to the PICU because of pneumonia. Because I am very susceptible to chest infections, almost every month I am admitted to the PICU.

At 10 months, I was diagnosed with CHOPS SYNDROME, the first and only case here in the United Arab Emirates. At present, I am rolling over and sitting with support. Since I am prone to aspirations, my feeding is only through the G-tube; that's why Pacifier is my happiness.

I also have hearing loss in both ears. I'm very excited whenever they put on my hearing aids because I love to hear my mama's voice. She always sings to me, and I'm so in love with her beautiful voice. I also love my dad because he always makes me happy. He takes care of me whenever Mama is at work. I love them both so much, and I'm so lucky to have them as my parents.

Thank you for reading!

ACCOMPLISHMENTS OF CHOPS FOUNDATION TILL NOW

CHOPS FOUNDATION SCIENTIFIC COMMITTEE

Manuela Mallemaci, founder of the foundation

A mother's wish to try and find ways to make life better and easier for her child.

SCIENTIFIC COMMITTEE

Dr. Ian Krantz, MD, attending physician in the Division of Genetics at Children's Hospital of Philadelphia, Director of the Roberts Individualized Medical Genetics Center, Director of the Center for Cornelia de Lange Syndrome and Related Diagnoses and the Genetics Residency/Fellowship Director. He holds a Distinguished Chair in the Department of Pediatrics.

Dr. Kosuke Izumi, MD and Assistant Professor of Pediatrics and member of the Division of Pediatric Genetics and Metabolism at UT Southwestern Medical Center, Dallas, Texas

Dr. Valentina Massa, professor in Applied Biology at the Department of Health Science, University of Milan, Italy.

Dr. Emanuela Scarano, medical director of Paediatrics – Rare Genetic Disease Division, Department of Woman and Children's Health, IRCSS Policlinico Sant'Orsola, Bologna, Italy

Dr. Katsuhiko Shirahige, researcher at Laboratory of Genome Structure and Function, Institute for Quantitative Biosciences, University of Tokyo; Karolinska Institutet, Dept. of Biosciences and Nutrition, Dept. of Cell and Molecular Biology,Biomedicum, Stockholm.

Dr. Eleonora Orlandini, pediatrician at the Rare Genetic Disease Division, Department of Woman and Children's Health, IRCSS Policlinico Sant'Orsola, Bologna, Italy

External consultants

Dr. Neil Hackett, genetic consultant for the Foundation and Research Professor in Genetic Medicine Weill Cornell Medical College, NY, US.

Funding for research is of utmost importance.
Our kids are too precious and not TOORARETOCARE.
We all care. There is power in teamwork.

CONFERENCE IN SIENNA - MAY 2023

Manuela writes:

Hello families,
some days ago our Foundation had a kick-off meeting with some members of our medical and scientific committee.
(If you want to know more about members of the board, just go here https://fondazionechopsets.com/en/pages/la-commissione-scientifica - Dr. Krantz is our president).

We concluded that before starting any therapeutic path we need to collect some more information, in order to better elucidate the underlying mechanism of CHOPS (dominant negative or haplo-insufficient?). In addition, this is extremely important in order to understand how many cell lines are important to develop: cell lines can be useful for designing a targeted therapy.

I would be very happy if you would share with me and the commission your kid genotype. A complete genetic report would be the best, but also only the type of specific mutation I think it is enough for the moment.

You can contact me anytime you want, I always answer also on FB messenger.

Urgent request to all parents to participate and complete:

Data Collection Survey at https://www.chopssyndromeglobal.org.data-collection.html

Family spreadsheet developed by Kayla Caceres

https://docs.google.com/spreadsheets/d/16bPcN6x0sujIi6qCLevnlUTNtm5rQfZd22edWEG0mQQ/edit

A medication spreadsheet will be out soon to be completed, prescribed medication (negatives and positives when using them).

SALVADOR

Hi, my name is Salvador Sandilands-Steyn. I have two moms, Elizly (Maps) and Rae-Dawn (Moomi). I was born at 38 weeks during an emergency C-section on 10 July 2021. The doctors struggled to get me out and had to use forceps, so I came out a little bruised. I didn't cry and I was a bit unresponsive. I was taken to the NICU because I had swallowed a lot of amniotic fluid. I had a tongue and a lip-tie, which they had to release and as soon as the geneticist saw me, he wanted to have some genetic tests done. I had a gastric nasal tube that I kept on pulling out, the nurses had to help me latch onto Moomi, which was unsuccessful, so they then tried the bottle. I gave them a big fright because with my first swallow attempt, I desaturated and aspirated. So, the next plan for me was a feeding tube directly into my stomach.

When they had to put in the PEG, they discovered that I had Laringomalacia. I had a funny way of breathing but that became a soothing sound to Moomi and Maps, because then they would know that I was still breathing. Moomi and Maps said, "HELL NO!" when the doctors suggested a tracheostomy.

We could go home after four weeks in hospital. I then got Sleep Apnoea with breathing pauses between 12 and 15 seconds at a time. Moomi and Maps had to ensure that I was always lying on my sides.

Then came the first big challenge whilst at home. I had to learn how to swallow. Moomi and Maps used a 0.1ml syringe and would drop one droplet of Rooibos tea on a dummy in the corner of my mouth. Gee, and did we have a couple of scares when I would choke on the droplet. Eventually, one drop became two drops and one syringe became two syringes. That doubled until I could eventually finish one 180ml of tea and 180ml of milk – and boy, did I love the taste!

Because I was drinking so well, my moms decided that I didn't need the PEG anymore and that they could take it out at 6 months. The hole where the PEG was didn't close and formed a sinus. This was my party trick – when I laughed, coughed or sneezed, the milk would shoot like a fountain. So, a plastic surgeon had to close the hole and for the first time in eight months, I could go onto my tummy. I didn't enjoy tummy time at all.

I kept on getting sick with flu or upper airway infections at least once a month. So, Moomi and Maps had to make sure I was not exposed to anyone who could possibly have the flu or a cold. Every time I would go to my music group called Wriggle and Rhyme, I would be sick within 48 hours.

The physio lady would come to our house and do some exercises with me, which made me very tired. It took a long time for me to roll from side to side and even longer to start moving my arms and legs in what looked like a leopard crawl. When I was 18 months old, I got a very bad skin rash and Moomi and Maps decided to take me to the paediatrician. Whilst we were there, the paediatrician said she might as well do my check-up. I had missed a lot of milestones and she asked Moomi and Maps if we had any feedback from Dr Bailey, the geneticist.

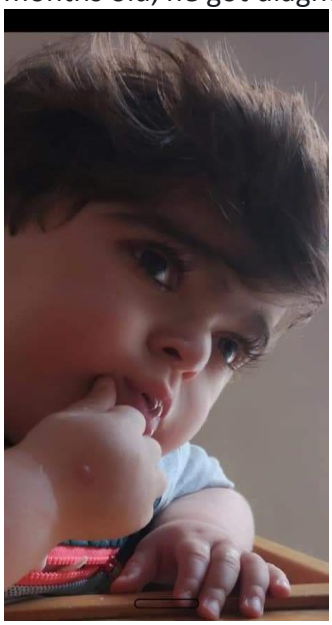
So Moomi and Maps decided to go and see Dr Bailey, where he mentioned that there was an AFF4 mutation and after looking at my limbs, my full head of blonde hair, my lashes (my girlfriends call me Sir Lash-a-lot) and my chubby cheeks, I finally became one of the very few members of the CHOPS family. My moms say that I'm a trooper, the friendliest boy with the bravest heart and cutest laugh. Although I can be a flirt with my special wink and gorgeous blue eyes and everyone adores my company.

Much love, Salva

MARIO

Hi, my name is Manuela and I am the mother of a very sweet two-year-old kid. At 5 months he was found to have multiple ventricular defects.

He had two cardiac surgeries, spending a total of 4 months of his life in a hospital. When he was 22 months old, he got diagnosis of a rare genetic disease.



My little boy, Mario has CHOPS syndrome, an almost unknown pathology that has only a few cases described in the medical literature and about 33 cases worldwide.

Mario is currently forced to use oxygen when he sleeps and if he gets sick, he has to use it 24 hours a day, not to mention the risk of possible serious pulmonary complications.

Mario has severe hypotonia; he's 2 years old, but he can't walk. Mario has moderate bilateral hearing loss. We also keep his eyesight under control; he has a severe myopia and a bilateral cataract.

He doesn't speak. It seems that language is a skill that CHOPS children are unlikely to develop.
Mario does physiotherapy and speech therapy when he is well, just to ensure that he can reach some important psychomotor milestones.

There are no research projects for CHOPS. Most doctors are not familiar with this disease. However, there is the possibility to find a cure! One of the therapeutic approaches relies on the drug screening. There are biotech companies with the right tools able to identify a drug capable of removing or at least alleviating the many symptoms that debilitate and limit the everyday life of CHOPS children.

As a mother, I have to act now, before the genetic mutation causes irreversible damage. I have started a GOFUNDME project https://gofund.me/c6c8f572 at the end of March.

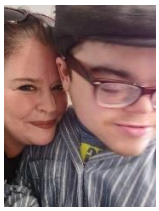
I am the President and co-Founder of a Foundation "Fondazione CHOPS Malattie Rare" https://fondazionechopsets.com/en. Our mission is to drive and accelerate a targeted therapy for CHOPS Syndrome. This foundation was established in May 2023 together with Luce's family.

The funds raised will be used for the program to identify a cure for CHOPS, WHICH WILL BENEFIT Mario and the other CHOPS children, rare like him.

WE HAVE MATURED SINCE 2019 ……..

Fashionable young Eider

Jacob (11) at the gym



Handsome Liam (22) with mom

Lovely Leta (25)

Alex (32) going for the big fish

AVERAGE MILESTONES FOR CHOPS KIDDIES and FREQUENTLY ASKED QUESTIONS ???????

FOR OUR NEXT NEWSLETTER OF OCTOBER/NOVEMBER, PLEASE GIVE FEEDBACK ON YOUR CHILD'S DEVELOPMENT AND SITUATION TO issteyn@yahoo.com

We know that each child has his/her own pace of growing and development, more so for kids with CHOPS.
However, it is of great assistance to new parents if they know about the delayed development that can be expected.

Holding head up steady, Sitting alone, Crawling, Walking, Speech, Comprehension, Puberty,

Sexuality and adulthood, Potty training, Medication prescribed (positives and negatives),

Emotional path of parents (guardians) and professional assistance, Familial support system.

AIMS FOR THE NEXT TWO YEARS - 2025

2024 - CONFERENCE 19 & 20 July

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

THE CHILDREN'S HOSPITAL OF PPHILADELPHIA.

Hopefully 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 RSVP

Lainey Moseley - **lwmoseley@comcast.net**

EXPAND RESEARCH

FUNDRAISING

MAKE IT BETTER FOR EACH AND EVERY ONE OF OUR CHILDREN

&

FAMILIES