Association for supporting children
with MECP2 duplication syndrome

NEWSLETTER

President's message

Dear members and supporters,

Two years ago, the acronym MDS was completely unknown to me. MECP2 duplication syndrome, a term that probably means nothing to you either. We are currently going through a crucial era in the treatment of genetic diseases, as well as cancers, infections, and other more common pathologies. The media has inundated us with information about CRISPR-Cas9, SiRNA, mRNA, ASO. But what do these enigmatic acronyms hide?

In reality, these terms represent a key to transforming the lives of some 300 million people affected by genetic diseases worldwide. They symbolize hope for children, parents, and families who aspire to see their loved ones grow up in a more serene environment. This prospect can sometimes raise fears and ethical questions, particularly concerning the modification of our DNA, and this is entirely understandable.

Nowadays, human life is marked by constant exponential growth in many areas. Our education system, largely based on acquired knowledge, must also reinvent itself to adapt to technological advances that can save lives. The syndrome we are particularly working on, MDS, is unfortunately progressive in most cases.

We have embarked on a race against time that pushes us to fight relentlessly to contribute to the colossal work already accomplished by other families over the past 10 years. Funding research projects, supporting families, and identifying new cases already represent a significant commitment made by our association but together, we can go even further.

Our children smile, make us smile, love, and are loved. They may not speak, but they communicate. Sometimes they don't walk, but they move forward. Our little and big ones are a source of inspiration, embodying resilience, and determination. When I feel melancholic, I look at my wife and children and think: "Everyone's life must have a meaning. Ours took a turn five years ago. Let's not waste time dreaming or hoping; what matters is action. So, let's go."

David Covini
President and co-founder

Themes

„Let's not waste time dreaming or hoping; what matters is action. So, let's go"

THE ASSOCIATION

Our story

Matteo was born on Labor Day in 2018. After three years of tireless struggle and feeling alone against helpless doctors, David and Caroline finally received the verdict on August 17, 2021: MECP2 Duplication Syndrome. The geneticist had never heard of it, and for good reason, as only 250 cases are registered worldwide.

They realized that support and communication for families were hindered by a significant lack of structure. Language barriers, complicated medical jargon, overwhelmed therapists, and incomprehensible procedures for applying for help were just some of the challenges they faced.

In May 2022, they decided to contact Austrian and German media to draw attention to the difficulties faced by parents of children with rare diseases and raise awareness of MECP2 Duplication Syndrome. The adventure began for this scientist couple from the pharmaceutical industry, originally from Switzerland and France, but living in Vienna for 10 years. Gradually, they connected with other parents with similar stories and were happy to find that some had already started paving the way for a solution: a drug.

After contacting most of the players in the field of MECP2 Duplication Syndrome, the Covini decided in the summer of 2022 to found their own organization, "Lasst uns MDS heilen – Dupmecp2" with the goal of accelerating research and more.

After reading an article about Matteo in the Kronenzeitung (Austrian newspaper), Gerald and Susanne, parents of Philipp who is also affected by MECP2 Duplication Syndrome, contacted the Covini family by email the same day. Soon after, the first personal meeting took place.

From left to right: David, Caroline, Ana, Borislav

Since then, the families have been in constant contact and exchange. When Susanne and Gerald learned that the Covini family was planning to found an association, they knew they wanted to be significantly involved. With 26-year-old Philipp, the Molnar family brings a lot of experience.

Shortly after, the Curic family from Upper Austria, consisting of Borislav, his wife Ana, and their two wonderful sons (Theo 6, Jan 2), discovered the Covini family's Instagram page. They immediately contacted them on the same day they received their youngest son Jan's diagnosis (also affected by MDS). Since then, they have achieved several milestones together as an association. The chance of a cure is realistic, which is why they decided to join the board as secretary. They are grateful not to have to walk this path alone.

From left to right: Susanne, Philipp, Gerald, Matteo, Caroline, David

Election of the board

We are excited to announce that the election for our Board Members and Auditors has been successfully completed! In light of the residential situation of our members, we opted for an online election, ensuring everyone had a chance to participate.

Application period:
We opened the floor for members to apply for a position until May 21, 2023.

Candidate Introduction:
We briefly introduced the candidates and their motivations to the members.

Election Timeline:
The election was conducted online from May 22 to June 1, 2023.

We are thrilled to announce that all applicants have been confirmed in their roles! We eagerly look forward to working together and achieving great things for our association.

David Covini
President

Gerald Molnar
Treasurer

Ana Curic
Secretary

Caroline Covini
Vice-president

Susanne Molnar
Deputy Treasurer

Borislav Curic
Deputy Secretary

Susanne Ertl
Auditor

Karl Svaton
Auditor

Missions of the association

The European association "Lasst uns MDS heilen – DupMECP2", based in Austria, advocates for children suffering from MECP2 duplication syndrome. Since its inception in 2022, the association has been working to raise funds to:

- **Promote and accelerate research to find a treatment**
- **Raise public and health professional awareness of this rare disorder**
- **Provide emotional and practical support to affected families**
- **Support the development of facilities adapted for disabled children**

In collaboration with other parents, friends, colleagues, doctors, scientists, and international associations, Lasst uns MDS heilen collects essential funds for research and provides support and information to families. Our belief is that unity is strength and that by working together, by joining everyone's efforts, we can have a significant impact on the lives of children affected by the syndrome.

„We are so grateful not to have to endure this journey alone."

Ana Curic, board member

Celebrating Milestones: Our 1-Year Achievements

On August 11, 2023, we celebrated the 1st anniversary of the association. We take this opportunity to review our actions over the past year:

- The association's board of directors, along with scientific experts, family members and friends, have brought together over 140 members from more than 10 countries. Together, we form a strong team and a global network committed to making a difference.

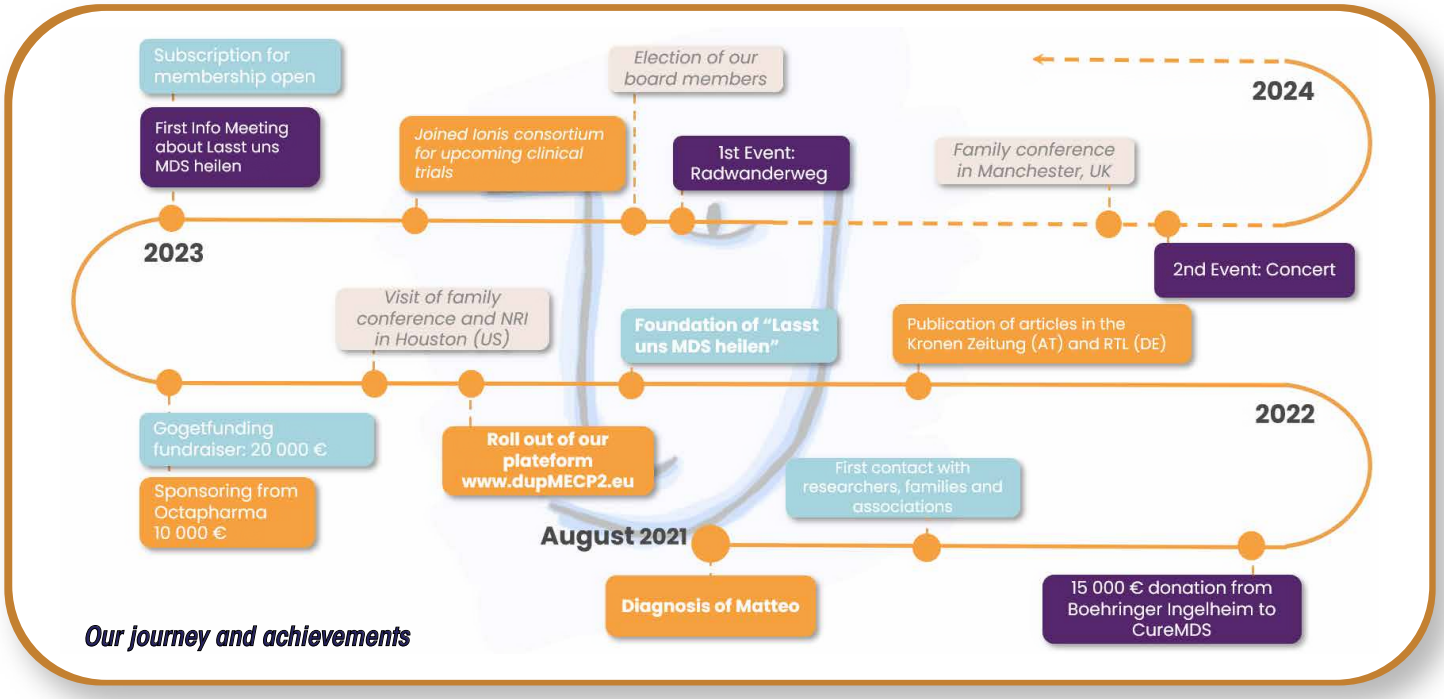
- A multilingual European platform was launched after months of effort and writing articles that closely address all aspects of MECP2 Duplication Syndrome:

www.dupmecp2.eu

- An initial event has taken place in the name of the association, and others are in preparation. Numerous donations have been received, and an ambitious project for massive fundraising is currently being developed and is expected to be launched by the end of the year.

- Since early 2023, Lasst uns MDS heilen is part of a worldwide consortium with Ionis Pharmaceuticals. This group is composed of representatives of several international associations dealing with MDS and is establishing upcoming clinical trials for the Antisense Oligonucleotides (ASOs).

Today more than a year ago, we are filled with emotion, and **we would like to express our immense gratitude to everyone who has been part of this journey.** Your support has been incredibly important.
Each step taken, each effort made, and each connection forged has contributed to the collective hope for a brighter future for our children.



Importance of becoming a member

In January 2023, Caroline and David, the visionary founders of "Lasst uns MDS heilen", stood alone. Today, we´re thrilled to count over 140 individuals in our growing family. But we´re not stopping there – our ambitions are even higher!
While we´re always delighted to have active members who can lend their support at various events and tasks, we understand and respect that not everyone may have time or capacity to do so.

BECOME A MEMBER

Every member, active or not, is a valued part of our team. Your membership also helps us demonstrate to authorities the strength of our support base, and thus, allows us to be eligible for public funds. We invite you to join us on this journey. A nominal membership fee of 10€ for individuals and 20€ for families, valid for the calendar year (up to 31st of December), is all it takes to be part of our mission. You also have the flexibility to opt for a 2 or 3 year membership.

Remember, everyone brings unique strengths to the table, and it's this collective power that makes us a formidable team. So, come, be part of our journey and let's make a difference together!

SCIENTIFIC PROGRESS

In the realm of rare genetic disorders, MECP2 duplication syndrome stands as a big challenge, affecting a limited number of individuals yet leaving an indelible mark on their lives. This complex disorder arises from the overexpression of the MECP2 gene, leading to a spectrum of neurological and developmental impairments that deteriorate with time.

In the past decades, researchers from 3 research groups have been working on innovative therapies for MECP2 duplication syndrome. Little by little, this research projects bring us closer to understanding and potentially treating this severe genetic disorder, giving hope to families and individuals dealing with its challenges.

ASO: A Breakthrough Discovery towards clinical trials

In 2015, a groundbreaking study conducted by Dr. D. Pehlivan and Pr. H. Zoghbi at the Neurological Research Institute (NRI, Houston, US), and Ionis Pharmaceuticals demonstrated that MECP2 duplication syndrome, could be reversed using Antisense Oligonucleotides (ASOs).

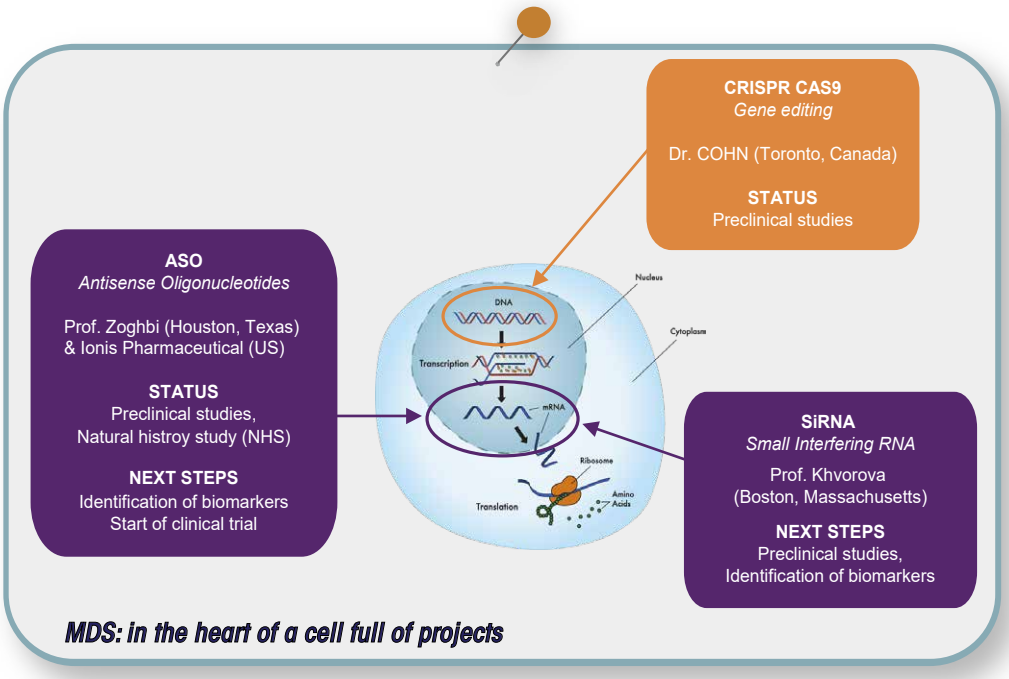
ASOs are short, synthetic pieces of DNA that can bind to the mRNA, thereby preventing the production of the corresponding protein. In this case, ASOs were designed to bind to the MECP2 mRNA, reducing the overproduction of the MeCP2 protein. The treatment not only stopped the progression of the syndrome but also reversed some symptoms, showing that the condition is not permanent. The successful use of ASOs in this study has paved the way for administration in humans, bringing us closer to a viable treatment option for this severe and progressive syndrome. **Complementary studies are currently underway towards clinical trials.**

The Power of siRNA and the Need for Funding

At the UMass RNA Therapeutics Institute (Boston, US), Dr. Anastasia Khvorova leads a promising project focused on using small interfering RNA (siRNA) technology to combat genetic disorders like MECP2 duplication syndrome. siRNA molecules silence specific genes by binding to and degrading their messenger RNA (mRNA), preventing protein production. The project has shown significant activity towards MECP2 mRNA, demonstrating siRNA's potential as a viable treatment option. However, **the project urgently requires $3 million in funding** to continue studies, refine the technology, and move closer to a cure.

The Power of CRISPR-Cas9

In Toronto, at the Sick Kids Hospital, Dr. Ronald Cohn is supporting a research group that is focusing on a revolutionary project involving CRISPR-Cas9 gene-editing technology. This group, led by Dr. Zhenya Ivakine has the potential to provide a treatment for genetic disorders, specifically targeting MECP2 duplicated gene. CRISPR-Cas9 works like molecular scissors, allowing scientists to alter, add, or remove specific parts of a DNA sequence. Although the project is currently less advanced than the ASO and siRNA projects, gene editing represents an innovative treatment that could potentially cure the disorder at its genetic root with a single administration.



Funding is crucial for advancing research and transform hope into reality.

MAKE A DONATION

More details about research projects in our next newsletter

EVENT

Bike tour in Gramatneusiedl

On June 18th, Gramatneusiedl (Lower Austria) hosted an event that showcased the spirit of generosity and community. As part of a bike tour organized by the city of Gramatneusiedl, our association "Lasst uns MDS heilen" was allowed to operate the refreshment stand.

Susanne and Gerald, esteemed board members, were at the heart of this circuit. They organized and managed the replenishment booth, where drinks and food were

available for a voluntary donation. Every single donation made during the event was dedicated to supporting the initiatives of the association. The booth, buzzing with activity, welcomed over a hundred visitors who showed their solidarity and support.

Matteo, a brave 5-year-old, and Philipp, a resilient 26-year-old, both living with MECP2 gene duplication syndrome, were present. Their inspiring presence shed light on the challenges faced by individuals affected by this rare condition and amplified the significance of the event's mission.

The success of this event was further magnified by the invaluable help of friends and supporters, who volunteered in organizing and operating the booth while proudly wearing the colors of the association.
We would also like to express our gratitude to the company Stiegl, who donated all the drinks! Furthermore, delicious pastries incl. coffee and delicious spreads were provided.

Reflecting on the event, Gerald says: "I am overwhelmed and infinitely grateful." Gerald is overwhelmed by the numerous volunteers who dedicated their time and energy, grateful for the vibrant and pleasant atmosphere that prevailed throughout, in awe of the significant number of visitors who joined, moved by the immense willingness to contribute and make a difference, and delighted by the visitors' curiosity and interest in learning more about the condition.

Susanne also shared her feelings, exclaiming, "What a great day. I couldn't and didn't want to hold back my tears of joy"

She expressed her heartfelt appreciation for everyone who made the event a resounding success for the association. Thinking back about the event, Susanne says "For me, this day was one of the most emotional since the beginning of our journey." She confesses "There is hardly anything worse than feeling hopeless and seeing your child suffer"

Their journey continues with renewed hope and determination for their son Philipp, knowing that they are not alone.
Susanne concludes: "Many thanks to all who made this day, such a good day for the association"

„What a great day. I couldn't and didn't want to hold back my tears of joy."
Susanne Molnar, board member

Thanks to the collective efforts of the organizers, volunteers, visitors and supportive sponsors, more than 1500 euros were raised for the association, showcasing the incredible power of unity and compassion when directed towards a worthy cause.

„I am overwehlmed and infinitely grateful"
Gerald Molnar, board member

We thank Leni, Barbara, Ingrid, Nina, Robert, Hubert, Gerlinde, Albert, Susanne, Andi-Bub, Emilian and Marie for the invaluable help.

Our association would like to thank again the company Stiegl for the generous donation of drinks. Furthermore, we would like to thank the insurance office, Klapa, for organising the event and allowing our organisation to join in. Thanks also goes to the Hirnich confectionery for providing the cakes. Many thanks also to Liebesblick for the beautiful material (T-shirts, bags, stickers). Last but not least a big thank you to dear Silvie, who sponsored our association's shirts!!!

PRESS

We are pleased to share the recent press coverage our first event has garnered. The event brought our community together to raise funds and awareness of MECP2 duplication syndrome in lower Austria. Two local newspapers have captured the key moments of the day, shedding light on its purpose and impact.

Our gratitude goes out to all the individuals who played a part in making the event a success, from volunteers to sponsors and participants.
We're grateful for the media's attention and the opportunity to communicate our goals to a wider audience. With this news coverage, we hope to build upon our achievements and continue fostering positive change for children with rare genetic disorders.

NÖN Woche 26/2023 — Meine Region — Region Himberg

GRAMATNEUSIEDL

Radeln für guten Zweck

Die Partnerschaft Gramatneusiedl organiserte einen Radwandertag zugunstern der Forschung an der seltenen Krankheit MECP2 Duplikation Syndrom. Etwa 150 Radler waren dabei.

MANUELA SCHWARZ

GRAMATNEUSIEDL Gleichzeitig mit dem Fest der Feuerwehr gab es in Gramatneusiedl vor kurzem einen Radwandertag für den guten Zweck, wo Start und Ziel am Hauptplatz der Feuerwehr stattfand. Zahlreiche RadlfahrerInnen stärkten sich anschließend mit hausgemachten Köstlichkeiten der Feuerwehr.

Der Radwandertag wurde von der Partnerschaft Gramatneusiedl organisiert. Der Verein unterstützt Kinder mit MECP2 Duplikation Syndrom. Dabei handelt es sich um eine seltene und fortschreitende neurogenetische Störung, an der der Gramatneusiedler Philipp Molnar leidet. Sein Vater Gerald Molnar, Vorstand des Vereins, und ein befreundetes Ehepaar Caroline und David Covini, die beide in der Forschung tätig sind, haben ebenfalls einen Sohn Matteo, der an MDS leidet. Gemeinsam versuchen sie, mit Spendengeldern wie etwa durch den Radwandertag, an dem rund 150 [Rad]ler teilnahmen, die For[schung] ...

▲ Philipp Molnar, Eltern Gerald und Susanne Molnar, Caroline und David Covini mit Sohn Matteo.

◀ Günther Klapa und Gabi Hertl beim Start und Ziel des Radwandertags

Fotos: Manuela Schwarz

GEMEINDERAT

Widerna übernimmt SP-Sitz von Nagy

Karin Widerna wurde als neue SPÖ-Gemeinderätin von Bürgermeister Paul Frühling (ÖVP) angelobt.

MOOSBRUNN Nach dem Ausscheiden von Daniel Nagy aus dem Gemeinderat nominierte die SPÖ Karin Widerna als neue Gemeinderätin. Nun wurde sie in der Gemeinderatssitzung von Bürgermeister Paul Frühling angelobt. Ebenso übernimmt Karin Widerna die Agenden im Schul- und Kindergartenausschuss. „Ich bedanke mich bei Daniel Nagy für sein konstruktives Engagement, darf Karin Widerna im Team herzlich willkommen heißen und freue mich auf gute Zusammenarbeit!", betonte Frühling im Anschluss an die Gemeinderatssitzung.

▲ Bürgermeister Paul Frühling mi[t] Karin Widerna und SPÖ-Ch[ef] ...

Article from NÖN

NACHBA[R]

Gramatneusiedl: Radfahren hilft

Beim Radwandertag sammelte Verein „Lasst uns MDS heilen" für kranke Kinder.

Article from Servus Nachbar

SAVE THE DATE

Benefit concert live & pur

Get ready for an unforgettable night of austropop music at our benefit concert featuring the band Live & Pur on November 25, 2023! Live & Pur, known for their dedication to social causes, are thrilled to lend their talents to support our association.
All members of the association thank the music band from the bottom of their hearts, especially Kurt Jansa, the original member of the band, who initiated this project and made it possible!
Kurt has known Phillip, who suffers from MECP2 duplication syndrome, since he was born, which gives this concert a very personal and emotional character.
Get ready to be amazed by their performance while enjoying tasty food and drinks.
But that's not all! We'll also be hosting a raffle and selling exclusive club merchandise (shirts, bags, etc.) with the assistance of Nina (Lieblingsblick) to raise even more funds for our cause. Remember, every cent of the proceeds goes directly to our association! Don't miss out on this fantastic opportunity to make a difference while having fun. Only 200 tickets are available. Secure your tickets now for just €15 in advance or €18 at the door.

We guarantee a night filled with good vibes and great memories. The entire team of our association eagerly awaits.

Important info about the concert

- Date: 25th of November 2023
- Location: Gemeindezentrum, Marie Jahoda Platz, 2440 Gramatneusiedl
- Entry: 18.00
- Start: 19.00
- Pre-sale: 06646117833, or office@dupmecp2.eu
- The tickets will be reserved after receipt of payment
  Erste Bank: AT 27 2011 1846 6994 4200

13th Austrian Congress for Rare Diseases

Topic: Hope. Looking to the future: Focus on gene therapies for rare diseases

The program (including gene therapy - "game changer" in rare diseases, gene therapy and novel therapies, advanced therapy, medicinal products, ATMPs, everything you wanted to know about gene therapy, etc...) offers both days interesting lectures to a broad audience. Lasst uns MDS heilen will attend the event to gain deeper insights into gene therapies and explore potential future treatment options for our children.

- Date: 6. and 7.10.2023
- Location: 1090 Wien, Josephinum

Family Meeting in Manchester: United for MECP2 Duplication Syndrome

Join us for the MDS Family Conference in Manchester on October 28, 2023! This exciting event (organized by mecp2duplication UK) aims to unite experts, physicians, affected individuals, and supporters to discuss the latest breakthroughs in MECP2 Duplication Syndrome research.
Representatives from Ionis Pharmaceuticals and the NRI (Houston) will be present to share their current work and future plans regarding ASO project. Additionally, several speakers will provide insights on managing various symptoms, and status of other research projects.
Our board members, Ana and Caroline, along with our medical expert, Prof. Gudrun Gröppel, will attend the conference and bring back valuable information to share with our community.

Don't miss this opportunity to connect, learn, and make a difference in the fight against MECP2 Duplication Syndrome!

- Date: October 28, 2023
- Location: Holiday Inn, Manchester, UK
- For more info: contactus@duplicationmecp2uk.org.uk

OUR HEROES

We introduce our heroes

Since our members are scattered all over the globe, it is difficult to meet them in person. That is why we would like to introduce you to an affected child, parent or member at the end of each newsletter.

Family Molnar

Gerald and Susanne live with their son Philipp in a single-family home located south of Vienna. They have been married for 37 years. Their daughter Susanne lives with her husband Albert near Lake Neusiedl in Burgenland. They are active in the association as treasurer and deputy treasurer. Through their involvement, they aim to contribute to the research for a treatment. Here, we would like to introduce you to their son Philipp.

From left to right: Susanne, Albert, Susanne, Gerald & Philipp Molnar

Philipp, 26-year old

This is our son Philipp Molnar. He was born in April 1997. The pregnancy was uncomplicated, the birth a bit difficult, but everything seemed to be fine... But reality soon caught up with us and we had to realize that Philipp has a disability.

This disability changed the life of the whole family! Despite the many handicaps, Philipp was an agile, happy and fun-loving youth until the onset of epilepsy.

He loved folk music, was enjoying every holiday and wanted to discover something new every day. He could go on long hikes without any problems and occasionally teased his older sister. Due to his drug-resistant epilepsy, it was only determined in 2021 that he had a rare genetic disease called MECP2 duplication syndrome.

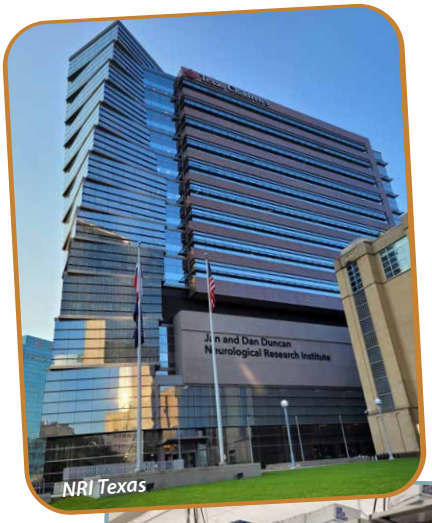
Philipp's constant companions are the symptoms that this gene disease brings with it. Such as lack of speech, impaired motor coordination, recurrent pneumonia, constipation and a drug-resistant epilepsy with multiple seizures every day.

Philipp and his family hope from the bottom of their hearts that one day his illness can be treated.

Susanne & Gerald

BACK TO THE CONFERENCE IN HOUSTON IN PICTURES

In September 2022, the family conference was held in Houston at the NRI. It provided an incredible opportunity for the participants to meet with passionate people dedicated to making a difference through advocacy and research.


NRI Texas


From left to right:
Dr. Pehlivan, Prof. Zoghbi,
David Covini, Ak Muharrem,
Dr. Suter


Peter, Ollie (CureMDS)
& David Covini


ASO laboratory in the NRI


Aron Schmidt (MECP2duplication foundation) & David Covini


David Covini
& Prof. Huda Zoghbi

OUR SPONSORS

We thank the sponsors that support our activities and the expansion of the association. All this would not be possible without them.

THANK YOU!

MEMBERSHIP

BECOME A MEMBER

Having more members is important for us! Support our missions and become a member of the association!

DONATION

PAYPAL

Bank transfer
Bank: Sparkasse Erste Bank
IBAN: AT27 2011 1846 6994 4200
BIC: GIBAATWWXXX

CONTACT

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