MENKES INTERNATIONAL ASSOCIATION

ANNUAL REPORT

2024

MAKING MENKES DISEASE HISTORY




"Who has health, has hope; and he who has hope, has everything."

Attributed to the Scottish historian and writer,
Thomas Carlyle.

This year's chosen quote speaks to both health and hope—a sentiment that resonates deeply with our work at the Menkes International Association *(MIa)*. Our mission has continued to strengthen year by year, but it's the hope we've been able to bring to children affected by Menkes disease and their families that stands as our greatest achievement.

Despite countless hours dedicated to networking, advocacy, and communication, it is our ability to provide real, life-saving treatment options to children affected by Menkes disease that sets us apart. The most significant of these efforts has been the continued expansion access to the experimental drug elesclomol-copper, alongside a tailored dosing plan and ongoing clinical support for severely ill children. Currently, we are seeing the number of children in the MIa Exceptional Treatment Program steadily grow, with five children receiving this life-changing treatment in 2025. The waiting list for those awaiting legal approval continues to expand across different countries, further underscoring the urgency of our work.

It is important to note that MIa's role is not to replace clinical trials and scientific research into Menkes disease, but to provide treatment during the interim for children affected by Menkes disease. Children affected by Menkes disease don't have time any to wait, and we maintain our commitment to never abandon any child who has received the necessary legal approval for treatment, particularly when early results are so promising. Elesclomol-copper is the first new medication for Menkes disease in over forty years—a breakthrough made possible by science and by MIa's partnership with Yusuf Hamied, philanthropist and founder of Cipla Therapeutics, who has generously provided the drug to MIa initially for Marco which could be used for other children affected by Menkes disease free of charge.

Our clinical team represents the world's leading Menkes disease specialists. In 2024, MIa's team worked tirelessly to ensure every child affected by Menkes disease has access to the best care possible MIa's Copperless Committee—an exceptional scientific body—continued to guide our efforts with deep expertise. We extend special thanks to the Spanish Medicines Agency *(AEMPS)* for their invaluable support. Thanks to our continued efforts and your continued support, we remain optimistic that an effective prevention and treatment options will resign Menkes disease to history.

With gratitude for your support,

Aurora Mateos
Founder and Director
Menkes International Association

TABLE OF CONTENTS

MIa 2024:

TEN KEY ACHIEVEMENTS

1. Menkes Patient Registry

In 2022, MIa proudly launched the first and only global Menkes Patient Registry, which continued to be expanded on 2024. This confidential, free online platform connects families, medical professionals, and researchers, fostering collaboration and accelerating the development of the development of Menkes treatment options. With 134 Menkes cases now registered across 25 countries, this registry is a unique and crucial tool to advance both clinical research and patient care. It plays an essential role in clinical trials, treatment planning, and improving the quality of life for Menkes patients worldwide. The MIa registry is not just a database—it's a vital resource for families seeking support and hope. Managed by a dedicated volunteer, we aim to continue to expand its capacity in the future.

2. Elesclomol-Copper Treatment

Since January 2021, MIa has facilitated access to elesclomol-copper treatment. The first child treated with this medication, Marco, continued to show remarkable improvements—he can walk, run, and speaks two languages. Inspired by Marco's progress, MIa expanded expanded this treatment to XX other children globally, working closely with local hospitals and authorities to ensure safe and effective administration.

3. Copperless Committee

MIa's Copperless Committee, made up of world-renowned experts, regularly meets to guide treatment decisions. They evaluate patient progress, modify treatment protocols as needed, and ensure that the highest standards of care are followed. This Committee plays a critical role in advancing scientific understanding and improving patient outcomes for Menkes disease.

4. The Core Team

The MIa Core Team, our pharma-clinical team, continued to oversee the follow-up care of patients receiving elesclomol-copper treatment. This multidisciplinary team supports local doctors and provides clinical advice. In 2024, we strengthened the team by appointing Dr. Elena Godoy as Coordinator of the Core Team and reinforcing our pharmacological expertise.

5. Global Collaboration, Scientific Conferences, and Advocacy

MIa continues to build relationships with regional and global health organizations, scientific communities, and advocacy groups. Our collaborative efforts were showcased at the prestigious Sorrento Copper Meeting in 2024, where the Copperless Committee shared the latest advancements in Menkes disease research and therapies.

MIa 2024:

TEN KEY ACHIEVEMENTS

6. Menkes and Other Rare Copper Diseases Meeting (Marco I)

In an unprecedented move, MIa organized the first-ever global conference focused on rare copper-related diseases. This milestone event, held alongside the Sorrento Copper Meeting, highlighted the urgent need for more research into copper metabolism and its role in rare diseases.

7. Annual Charity Gala

MIa's charity gala in Málaga, Spain, has become a highly anticipated annual event, with tickets for the 2024 Gala event selling out within 48 hours and a waiting list of over 100 people. The event continues to raise awareness about MIa and vital resources to advance our mission.

8. Christmas Charity Event

In 2024, MIa hosted its first-ever Christmas charity event, where children played a central role in spreading joy and raising awareness about Menkes disease.

9. Patients' Personal Impact

MIa's impact is not just about research—it's about life-changing, tangible results in the lives of children with Menkes disease. Marco's story and the stories of other children undergoing treatment with elesclomol-copper highlight the profound difference this treatment is having on their condition and quality of life. Families who once had no hope now see their children with Menkes disease achieving milestones they thought were impossible.

10. Financial Accountability

MIa upholds the highest standards of financial transparency and accountability in the use of resources. The majority of these resources are allocated to treatment, family support, and advocacy initiatives. The support of volunteers allows us to direct as much funding as possible toward helping Menkes children and their families.

1.Menkes Patient Registry

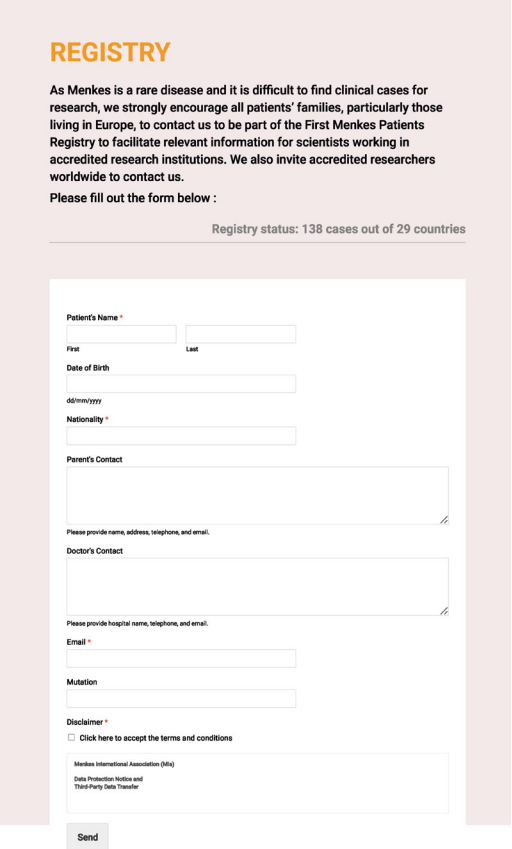
In 2022, the Menkes International Association (MIa) launched the first and only global Menkes Patient Registry. This innovative, free online platform bridges the gap between families of children affected by Menkes disease and leading medical and scientific research teams at accredited institutions. The registry not only provides families with access to cutting-edge research but also facilitates collaboration between researchers to identify more effective treatments and, ultimately, a cure for Menkes disease.

The significance of the Menkes Patient Registry cannot be overstated. As noted by Orphanet, patient registries are vital tools in rare disease research. They enable the sharing of critical data, which is fundamental to clinical research. By aggregating patient data, we are able to better design clinical trials, evaluate the effectiveness of treatments, and enhance patient care for children with Menkes diease. For rare diseases like Menkes, where treatment options are extremely limited, this data-driven approach is essential for advancing our scientific understanding and accelerating the development of new therapies.

As of the end of 2024, the Menkes Patient Registry had enrolled 134 children from 25 countries. This growing global network is much more than a simple database—it is a lifeline for families seeking support and hope. It fosters a strong community of patients, researchers, and clinicians who are united in the fight against Menkes disease.

Currently, the registry is managed by a dedicated volunteer who works part-time on weekends. Looking ahead, MIa is actively seeking funding to hire additional staff, ensuring the continued expansion and effectiveness of this invaluable resource. Our goal is to guarantee that the registry remains a free, easy to access and trusted support system for families affected by Menkes disease around the world.

> For more information, please visit the Menkes Patient Registry:



REGISTRY

As Menkes is a rare disease and it is difficult to find clinical cases for research, we strongly encourage all patients' families, particularly those living in Europe, to contact us to be part of the First Menkes Patients Registry to facilitate relevant information for scientists working in accredited research institutions. We also invite accredited researchers worldwide to contact us.

Please fill out the form below :

Registry status: 138 cases out of 29 countries

Patient's Name *
First
Last
Date of Birth
dd/mm/yyyy
Nationality *
Parent's Contact
Please provide name, address, telephone, and email.
Doctor's Contact
Please provide hospital name, telephone, and email.
Email *
Mutation
Disclaimer *
Click here to accept the terms and conditions
Menkes International Association (MIa)
Data Protection Notice and Third-Party Data Transfer
Send

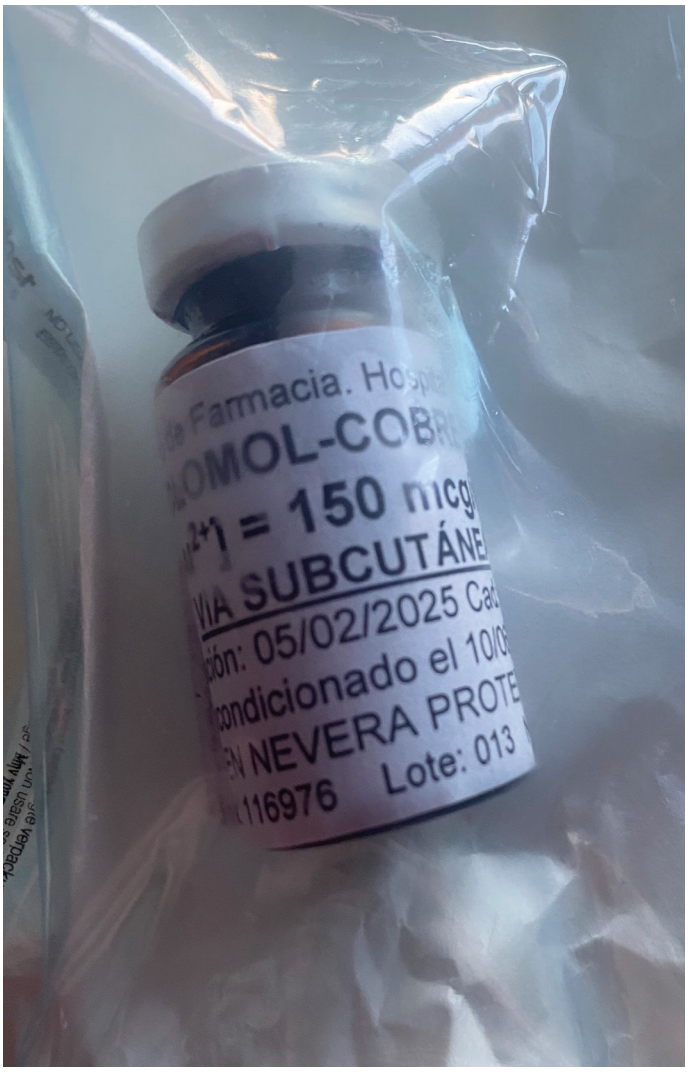
Funds raised: *N/A*
Funds spent: *N/A*

2. Elesclomol-Copper Treatments for Menkes Patients

2.1 Status of the Treatments

Since January 2021, the Menkes International Association *(MIa)* has been at the forefront of expanding access to the groundbreaking elesclomol-copper treatment for children affected by Menkes disease across the globe. This treatment, which was first successfully tested on a young boy named Marco, has since brought renewed hope to many children suffering from this devastating condition.

Following Marco's remarkable progress, MIa initially made the treatment available to other children in Spain. We are now actively working towards expanding access to this life-saving treatment in additional countries. To facilitate this, we are collaborating with national medical teams to obtain the necessary authorizations. As of the end of 2024, five children are receiving this treatment in Spain While we await the commencement of formal clinical trials, this therapy represents the only viable treatment for these life-threatening cases of Menkes disease. To learn more, please read the patient's story *(section 9)*.



CORE TEAM

Looking forward, MIa remains optimistic about the future. With the continued support and engagement of our partners, donors, and the broader community, we are committed to expanding access to this treatment, accelerating ongoing research, and ultimately making Menkes disease a disease of the past.

2.2 Expanding Access and Treatment Protocol

One of MIa's most significant contributions to the fight against Menkes disease is our ability to expand access to treatment—something that no other organization currently offers for children with this rare condition. Marco's extraordinary progress served as a catalyst for MIa to plan to extend this treatment to more children worldwide. Our plans to expand access to treatment is rigorous and follows a strict protocol:

1. A request must come directly from the patient's medical team.
2. The patient must receive treatment at their local hospital.
3. The family must provide informed consent to work with MIa.
4. All parties involved—including the medical team, the family, and MIa—must agree to collaborate for the child's benefit.
5. National drug authorities must grant approval for the use of treatment.

Although MIa's work does not replace formal clinical trials, it has proven to be a vital lifeline for the children involved. Each child receiving treatment has demonstrated notable improvements in quality of life, providing their families with renewed hope for the future. MIa remains steadfast in its commitment to expanding this initiative and ensuring that no child with Menkes disease in need is left waiting for treatment.

Funds Raised: €13,000
Funds Spent *(including Core services and travel expenses)*: €43,002

3. Copperless Committee Activities

The Copperless Committee is an esteemed international team of experts from diverse fields, dedicated to providing critical support to patients with Menkes disease who are receiving the elesclomol-copper *(ES-Cu)* treatment. This multidisciplinary group convenes monthly to offer specialized advice, ensuring that treatment decisions are informed by the latest scientific advancements.

During these meetings, the committee reviews the progress of each patient, adjusts treatment dosages as necessary, and evaluates whether to continue or modify the treatment plans. Additionally, the committee assesses new patients eligible for the therapy and remains up-to-date on the latest scientific insights into copper metabolism, which is vital for optimizing patient outcomes.

The committee's work plays an essential role in ensuring that every treatment decision for Menkes patients is evidence-based, drawing from the most current clinical and research data. By providing expert guidance, the Copperless Committee helps refine and enhance treatment strategies for children fighting this rare disease.

On December 1st, 2024, the Copperless Committee approved its Rules of Procedure, which can be accessed here:

Copperless Committee Rules of Procedure

2024 Sorrento Copper Meeting

In 2024, the Copperless Committee held an in-person meeting during the Sorrento Copper Meeting, an annual international event that focuses on the genetic, molecular, and biochemical mechanisms related to copper biology, as well as its role in diseases like Menkes. The meeting serves as a platform for both basic and translational scientists, as well as clinicians, to share emerging concepts and breakthroughs in the copper field. The committee's involvement in this event was significant, with five members delivering plenary lectures. Notably, three of these sessions focused specifically on the exceptional treatment protocol for elesclomol-copper.

The 2024 Sorrento Copper Meeting was a pivotal event for advancing knowledge in the copper field, featuring two keynote lectures and 36 plenary lectures. This collaborative platform continues to contribute immensely to the development of treatments and therapeutic strategies for copper metabolism disorders, including Menkes disease.

For more details on the conference and the role of the Copperless Committee, please visit the following links:

- 2024 Copper Meeting Website
- Copperless Committee Overview

Funds Spent: €7,685.97

4. The Core Team

The Core Team is dedicated to overseeing the follow-up care of patients receiving the elesclomol-copper treatment under an exceptional treatment protocol. This team provides vital support to local medical teams and offers clinical management advice, ensuring the best possible care for Menkes patients. Recently, the team has been restructured to further strengthen its pharmacological expertise. As per the procedural guidelines approved on December 1st, 2024, a new Coordinator was elected by the Copperless Committee. Dr. Elena Godoy has succeeded Dr. Francesc Palau of Sant Joan de Déu in this role. Together with a multidisciplinary group of medical and pharmaceutical professionals, the Core fosters seamless communication among families, clinicians, and researchers, ensuring the highest quality of care for Menkes patients. The CORE also provides advisory support to physicians and pharmacists at an international level, facilitating access to copper histidinate and assisting in the clinical care and follow-up of patients affected by Menkes disease.

The Current Members of the Core Team Include:

- **Dr. Elena Godoy** *(Coordinator)*, Pediatrician and Clinical Advisor, Pediatric Complex Chronic Patient and Palliative Care Unit, Hospital Regional Universitario de Málaga, Spain
- **Dr. Francesc Palau**, Geneticist, Hospital Sant Joan de Déu, and NeuroGene; Senior Investigator, SJD Research Institute and CIBERER
- **Ms. Rosa Marques**, Neurological Pediatric Physiotherapist and Head of Clinical Pediatric Physiotherapy Teams, Dulce Nombre de María Psychopedagogic Institute, Málaga, Spain
- **Dr. Stephanie Lotz-Esquivel**, Clinical Investigator, Institut de Reçerca Sant Joan de Déu, Barcelona, Spain
- **Dr. Miquel Villaronga**, Hospital Pharmacist *(retired)*, Hospital Sant Joan de Déu, Barcelona, Spain
- **Dr. Ángela Pieras**, Hospital Pharmacist *(Consultant)*, Hospital Sant Joan de Déu, Barcelona, Spain
- **Dr. Rubén Varela**, Hospital Pharmacist, Complejo Hospitalario de León, Spain
- **Dr. Mónica Sáez**, Hospital Pharmacist, Complejo Hospitalario de León, Spain
- **Dr. Aquilina Jiménez González**, Pediatrician, Department of Pediactrics Neurology, Complejo Hospitalario de León, Spain
- **Dr. Aurora Mateos**, Core Secretariat *(also MIa Founder and Director, Málaga, Spain)*

Operational Activities of the Core Team

The Core meets on a weekly basis to monitor patient progress, assess treatment response and the occurrence of adverse events, and assess and plan the implementation of new therapies. While its focus is primarily on children with Menkes disease in Spain, the team's activities have expanded internationally in 2024. Virtual meetings with doctors and families from countries such as Germany, France, and the UK have increased, further establishing the Core as a unique global reference for pharmaco-clinical expertise in Menkes disease. Additionally, the Core has worked in close collaboration with Spain's Medicines and Medical Devices Agency *(AEMPS)* to review and discuss treatment options for Menkes diease.

Meetings and Collaborations in 2024:

Core Team Meetings:

- February 1st, 2024
- February 15th, 2024
- March 7th, 2024
- April 8th, 2024
- April 18th, 2024
- April 28th, 2024
- September 17th, 2024 *(Sorrento)*
- October 7th, 2024
- October 14th, 2024
- November 18th, 2024

Copper(less) Committee Meetings:

- February 9th, 2024
- April 2nd, 2024
- May 9th, 2024
- July 4th, 2024
- September 18th, 2024 *(Sorrento)*
- November 7th, 2024
- December 12th, 2024

Pharmacology Teams Meetings:

- February 7th, 2024 *(Hospital Sant Joan de Déu)*
- February 21st, 2024 *(Hospital Sant Joan de Déu + Hospital Universitario de León)*
- June 26th, 2024 *(Hospital Sant Joan de Déu)*
- July 15th, 2024 *(Pharmacokinetics)*
- October 11th, 2024 *(Hospital Sant Joan de Déu + Hospital Universitario de León)*

The Bayley Scale Meetings:

- January 18th, 2024 *(Inés Medina)*
- February 6th, 2024 *(Anna Aguilar)*
- February 22nd, 2024 *(Inés Medina)*
- March 12th, 2024 *(Inés Medina + Anna Aguilar)*
- May 29th, 2024 *(Julita Medina)*

Core + MIa Meetings:

- January 11th, 2024
- April 26th, 2024
- June 26th, 2024
- July 18th, 2024

Patient Visits:

- La Línea de la Concepción *(MNK4)* - January 30th, 2024
- León *(MNK2)* – February 8-9th, 2024
- HSJD Barcelona *(German patient)* – February 13th, 2024
- Sevilla *(MNK3)* – March 17-18th, 2024
- HSJD Barcelona *(French patient and MNK6)* – April 28-29th, 2024
- HSJD Barcelona *(MNK5)* – May 30th, 2024
- León (MNK2) – September 28-30th, 2024
- HSJD Barcelona *(MNK1)* – October 1st, 2024
- Lanzarote *(MNK5)* – November 22-27th, 2024

POther Meetings::

- Menkes Early Diagnosis – January 5th, 2024
- Anatomic Pathology – January 22nd, 2024
- Copper Team from Murcia - January 30th, 2024
- Menkes Unit *(Ángeles García-Cazorla)* – February 21st, 2024
- Named Patient Data Sharing *(Engrail)* – March 19th, 2024
- Joana Claverol *(Clinical Trials SJD)* – April 17th, 2024
- Joana Claverol *(Clinical Trials SJD)* – May 2nd, 2024
- Menkes Unit *(Ángeles García-Cazorla)* – May 16th, 2024
- Spain's Medicines and Medical Devices Agency *(AEMPS)* – July 17th, 2024 *(Milena Peraita)*
- Joana Claverol *(Clinical Trials SJD)* – August 29th, 2024

Meetings and Collaborations in 2024:

Potential Patient Meetings

- February 6th, 2024 *(Venezuelan patient living in Spain)*
- February 29th, 2024 *(Colombian patient)*
- March 12th, 2024 *(German patient)*
- April 17th, 2024 *(Dr. Karin Tuschl – United Kingdom)*
- July 1st, 2024 *(Dr. Ares Sánchez – Valencia, Spain)*
- August 20th, 2024 *(Dr. Eduardo García – Lanzarote, Spain)*
- October 8th, 2024 *(Dr. Alisson Caldwell – USA)*
- October 10th, 2024 *(Dr. Ares Sánchez and Dr. Patricia Smeyers – Valencia, Spain)*
- October 14th, 2024 *(Dr. Thorsten Marquardt – Germany)*
- October 30th, 2024 *(Dr. Adriana Ulate – Costa Rica)*
- October 30th, 2024 *(Dr, Chiang, Colby, U.S.A)*
- November 5th, 2024 *(Dr. Birute Skerliene – Lithuania)*

Funds Raised: €13,000
Funds Spent: €43,002 *(including Core services and travel expenses)*

5. Global Collaboration, Scientific Conferences, Advocacy & Communications

5.1 EURORDIS

MIa is honored to be a full member of **EURORDIS-Rare Diseases Europe** alliance since September 2022. EURORDIS is a non-profit alliance comprising over 1,000 organizations dedicated to improving the lives of more than 300 million people globally affected by rare diseases. As a member, MIa now has the privilege of voting at the EURORDIS General Assembly, enabling the organization to actively participate in shaping the future of rare disease advocacy across Europe and beyond.

In addition to its membership in EURORDIS, MIa is committed to strengthening Menkes disease advocacy by collaborating with national organizations around the world. Notably, MIa maintains close ties with **Angelli per la Vita** and **Amici di Leonardo** in Italy, as well as various South American associations, all of which contribute to raising awareness and advancing research efforts for Menkes disease.

For more information, visit: EURORDIS Website

5.2 Scientific Conferences

MIa actively participates in key international conferences that are relevant to the field of rare diseases, particularly those focused on Menkes disease and copper metabolism disorders. However, due to resource constraints, MIa's participation in conferences in 2024 was limited.

The 2024 Copper Meeting in Sorrento was one of the key events attended by MIa, where the **Copperless Committee** played a pivotal role in advancing scientific discussions on copper-related therapies for Menkes disease *(see Section Four)*. This conference remains an essential platform for collaboration and the sharing of research findings within the global scientific community.

Funds Spent: €100 per year for membership

Menkes International Association - Aurora Mateos.

5.3 MIa Website

Launched: October 2021 – Ongoing

In 2021, MIa launched its official website, which has since become an essential resource for connecting families, researchers, and supporters worldwide with information and support related to Menkes disease. The website serves as a unique, central hub for the latest news, updates, and resources related to Menkes disease, providing a platform for advocacy and engagement, playing a crucial role in MIa's outreach and communication efforts, ensuring that the global community remains informed about the progress being made in the fight against Menkes disease.

The design and ongoing maintenance of the website have been generously supported by **Texas A&M University**, with in-kind contributions from **Jim Sacchettini**, Head of the Laboratory, and technical management provided by **Sid Rath** and **Saswati Panda**. **Aurora Mateos**, a member of the MIa Board, oversees regular updates of the website's content, ensuring it remains current and relevant to the needs of the Menkes community.

In addition to providing vital information on Menkes disease and MIa's initiatives, the website also facilitates donations, volunteer opportunities, and collaboration with researchers and medical professionals worldwide.

For more information, visit:
Menkes International Website

Amount Paid: €535.88 (cost of web hosting via Bluehost)

6. Menkes and Other Rare Copper Diseases Meeting (Marco I)

The **1st International Meeting on Menkes Disease** and **Other Rare Copper Diseases** was successfully held in Málaga on **September 12-13, 2024**. Organized by **Menkes International**, this groundbreaking event brought together leading medical specialists, researchers, and patient representatives from around the world. In addition to the in-person participation, the meeting was also broadcast online, making it accessible to a global audience.

The primary objectives of the meeting were to:

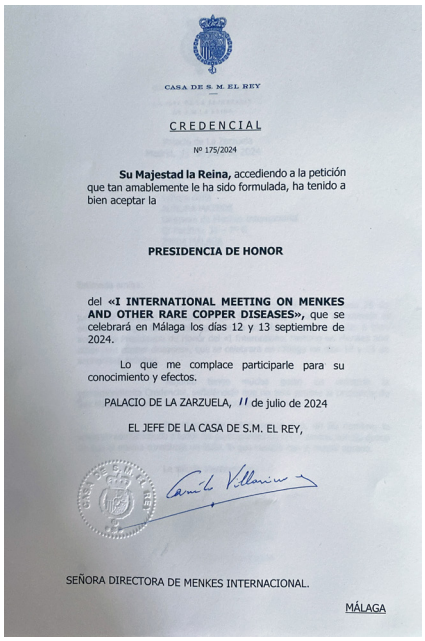
- Provide a platform for discussion and knowledge exchange on Menkes disease and other rare copper diseases.
- Foster collaboration and connections between physicians, researchers, patients, and their families.
- Promote awareness of these rare diseases, emphasizing the importance of early diagnosis and treatment.
- Strengthen the international community of professionals and advocates dedicated to advancing research and support for rare copper diseases.

Funds raised *(conference fees)*: 650 euros

Conference sponsorship by Engrail Therapeutics: €80,000 *(unrestricted grant which was also spent in exceptional treatments)*

Expenses: €31,156

The meeting concluded with the MIa charity gala, which successfully raised funds to support research and patient care initiatives.



CASA DE S. M. EL REY

CREDENCIAL

Nº 175/2024

Su Majestad la Reina, accediendo a la petición que tan amablemente le ha sido formulada, ha tenido a bien aceptar la

PRESIDENCIA DE HONOR

del «**I INTERNATIONAL MEETING ON MENKES AND OTHER RARE COPPER DISEASES**», que se celebrará en Málaga los días 12 y 13 septiembre de 2024.

Lo que me complace participarle para su conocimiento y efectos.

PALACIO DE LA ZARZUELA, 11 de julio de 2024

EL JEFE DE LA CASA DE S.M. EL REY,

SEÑORA DIRECTORA DE MENKES INTERNACIONAL.

MÁLAGA

We were honored to have the Queen of Spain as the President of Honour of the meeting , a recognition formalized through credential 175/2024. This support highlighted the significance of the meeting and the ongoing commitment to advancing research and care for Menkes disease and other rare copper diseases.

For more details, visit:
Menkes International Meeting Website

Event repap video:
https://www.youtube.com/watch?v=88xt8eBuMJM

7. MIa Charity Gala, September 2024

On **September 13, 2024**, Menkes International Association (MIa) hosted its **second charity fundraiser** at the prestigious **Hotel Miramar** in Malaga, Spain, one of the most luxurious hotels in the world. The event gathered **120 dedicated supporters**, each purchasing charity tickets for the cost **€100**. In addition to ticket sales, several generous donations were made during the evening, raising both awareness and crucial funds to further MIa's mission in the fight against Menkes disease.

The gala brought together key public figures, advocates, and philanthropists in support of MIa's cause. **Dr. Yusuf Hamied**, a prominent philanthropist, highlighted the critical role of copper in the treatment of Menkes disease. **Aurora Mateos** and **Denis Broun** represented MIa's leadership team, while **Dr. Elena Godoy** spoke on behalf of The Core, and **Nina Horn** represented the Copperless Committee.
The event's success was further amplified by the participation of six renowned local chefs, three of whom hold Michelin stars. These distinguished chefs included:

- **Diego Gallegos** *(Soho Restaurant)*
- **Diego René** *(Restaurant Beluga)*
- **Dani Carnero** *(Restaurant Kaleja)*
- **Kisko García** *(Restaurant Choco)*
- **Pablo Molina** *(Restaurant Matiz)*
- **Diego Nicas** *(Restaurant Príncipe de Asturias)*

In support of the gala, local companies generously donated the ingredients for the chefs to use in preparing the evening's culinary delights.

The event garnered significant public and media attention, with coverage from several prominent media outlets, helping to raise awareness about MIa's ongoing efforts to support families affected by Menkes disease.

Event recap video:
https://www.youtube.com/watch?v=5vTK5KOef1c

Total Funds Raised: €19,570
Expenses: €11,180

8. MIa Christmas Charity Event

The MIa Christmas Charity Event took place at the historic “Cofradía de Los Estudiantes” in Málaga, Spain, offering a festive and vibrant atmosphere for all attendees. The event, known as the tardeo navideño, featured a variety of activities, including live musical performances, games, and auctions.

The early part of the event was dedicated to families, with children taking center stage as they enjoyed the festivities. As the evening progressed, the atmosphere shifted, and the event became a lively gathering for adults. The celebration took place in front of the iconic Málaga Roman Theatre, creating a picturesque backdrop for this special occasion.

This event marked the beginning of the holiday season for MIa’s supporters, who gathered to celebrate and raise funds for the organization’s mission.

Funds Raised: €2,280
Funds spent: €1,090

9. Highlighting MIa's Personal Impact on Children with Menkes Disease

At Menkes International Association (MIa), our mission extends beyond advancing medical research. We are committed to transforming lives, as demonstrated by the inspiring stories of Marco and other children receiving the groundbreaking elesclomol-copper treatment. These success stories underscore the profound impact of hope, resilience, and the life-saving potential of scientific innovation. Families who once felt helpless are now witnessing their children with Menkes disease grow, thrive, and achieve milestones that were once thought impossible.

Marco's Story: A Beacon of Hope

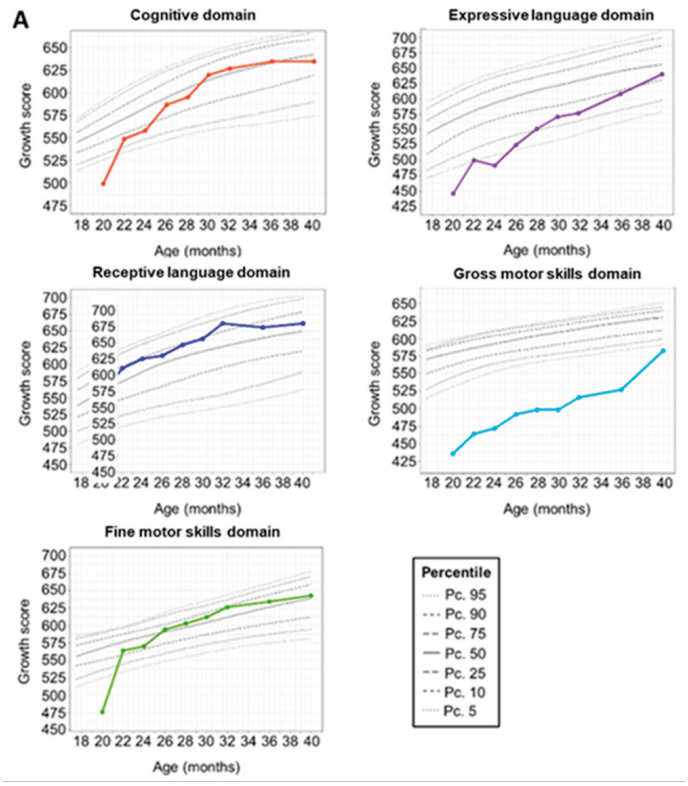
Marco's journey is nothing short of extraordinary. Once limited by the devastating effects of Menkes disease, Marco's progress has become a beacon of hope for families affected by Menkes around the world. After undergoing treatment with elesclomol-copper, Marco has learned to walk, run, and communicate—achievements that seemed impossible for a child with Menkes disease. As the child of MIa's founders, Marco's remarkable recovery is a public testament to the transformative power of innovative treatment. His progress continues to inspire hope for the future of all children affected by Menkes disease and serves as a symbol of the positive change MIa is helping to create.

More Patient Stories

Patient 1: A Life Transformed

Diagnosed with Menkes disease shortly after birth, patient MNK1 began treatment with elesclomol-copper in early 2022. Now four years old and almost after 3 years of treatment, MNK1 has made remarkable neurodevelopmental progress. He can walk steadily, run, and climb stairs—milestones once considered impossible for children with classical Menkes disease. His language skills are on track for his age, and he has even begun reading and writing. Despite facing other health challenges, including a pre-existing lung condition, the overall improvement in his quality of life has been life-changing thanks to this treatment.

Bayley Scale scores representing all five domains assessed at various ages (months).



Patient 2: Making Strides

Diagnosed prenatally in a family history of Menkes disease, patient MNK2 started Elesclomol-Copper treatment in 2023 with four months of age. Within just eight months, this two-year-old has made significant progress, especially in motor skills and cognitive development. Patient MNK2 can now walk independently and climb stairs, although some challenges remain. The development in motor control is a major achievement for this young patient.

Bayley Scale scores representing all five domains assessed at various ages (months).

Patient two is demonstrating more rapid motor development compared to patient one, achieving motor milestones at an earlier stage.

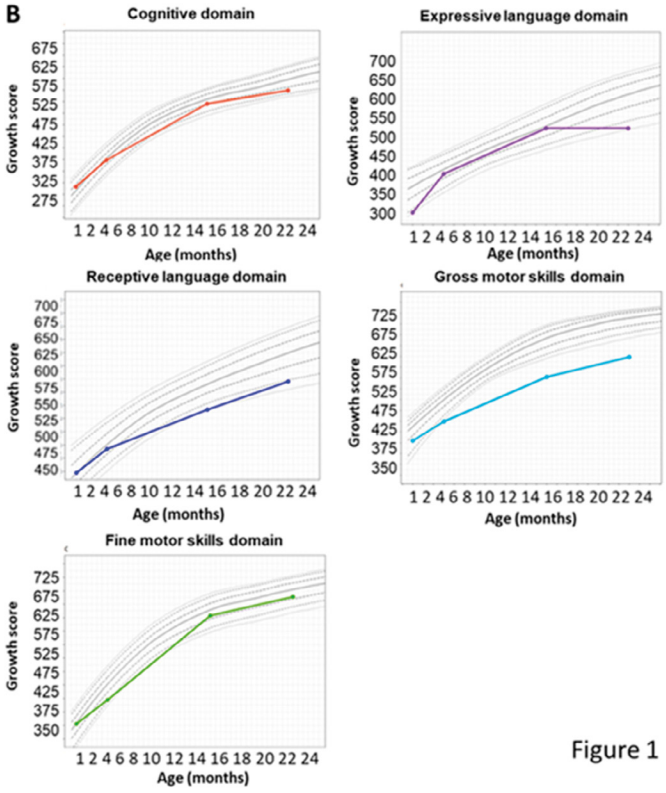


Figure 1

Patient 3: Overcoming Seizures

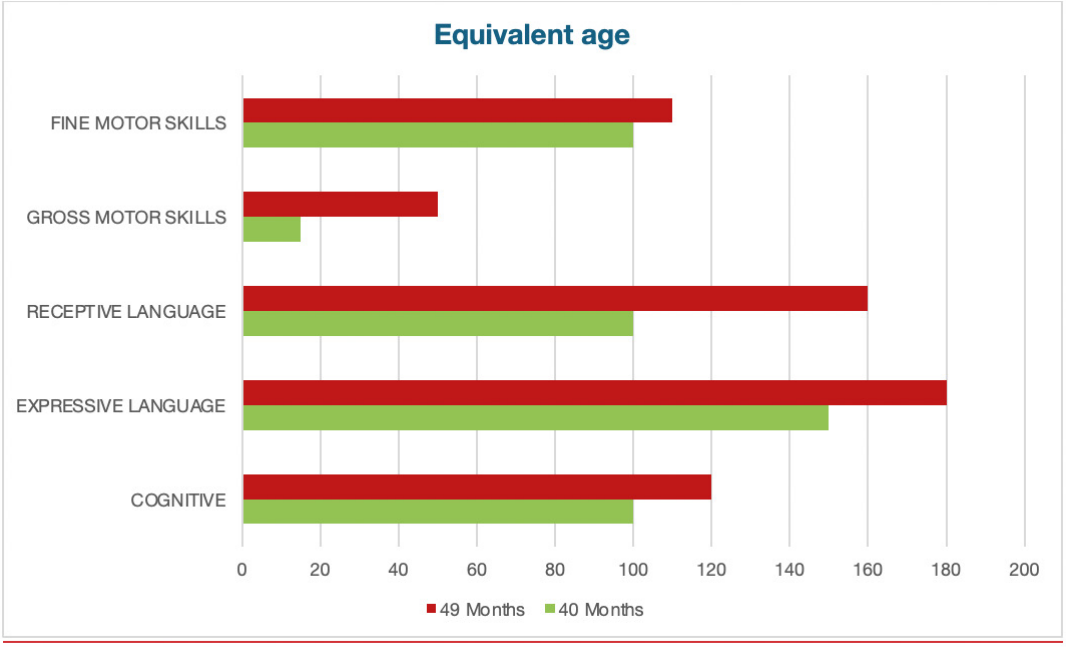
Diagnosed later than others, patient MNK3 has been receiving treatment for one year and six months, he was 15 months all when he started Elesclomol-Copper. Despite facing severe developmental delays and west syndrome, he has made modest yet meaningful gains in muscle control and engagement with his surroundings, including babbling. Regarding the occurrence of seizures, after 20 weeks of treatment a reduction in seizure frequency and improved control of spasms were observed, which has been maintained to date. Progress may be slow, but the quality-of-life improvements are evident for the family.

Equivalent age (months) across all five domains assessed through the Bayley Scale.

Patient 4: A Brighter Future

Patient MNK4, who began treatment in September 2023, was initially affected by severe neuro-developmental delays and was non-verbal. Since starting treatment, he has shown gradual improvements in muscle tone and communication. Early indicators suggest a promising outlook for his development.

Equivalent age (months) across all five domains assessed through the Bayley Scale.



Patient 5: Our Older Copper Warrior

Starting treatment at the age of 22 years, patient 5 received Elesclomol-Copper since November 2024; although the patient has been on treatment for a short time, an improvement in his behavior has been observed . He no longer experiences seizuresand has acquired new skills. His specific mutation allows him to produce some functional protein, which has contributed to his survival as one of the oldest patients with Menkes disease. His journey demonstrates the potential benefits of treatment even in later stages of the disease.

Looking Forward

While MIa does not conduct clinical trials, our work has proven to be a lifeline for these children with Menkes disease and their families. Each patient's progress demonstrates the profound improvements in quality of life that can be achieved with timely interventions. MIa is committed to expanding access to this life-saving treatment and ensuring that no child with Menkes disease is left without the care they need.

For more details on Marco's journey, as well as media coverage, follow the links below:

- Marco, la esperanza de niños con Menkes (RTVE)
- Un tratamiento experimental para Marco, el niño malagueño afectado por el síndrome de Menkes (CanalSur)
- La evolución de Marco abre el camino a otros niños con la enfermedad de Menkes (EFE)
- La proeza de Marco, el niño sin cobre | Diario Sur
- Marco, el niño con la enfermedad de Menkes que sobrevive gracias a un tratamiento pionero en España (ABC)
- The fight for Marco, the boy with no copper (Sur in English)
- El pequeño Marco consigue caminar gracias a un tratamiento pionero en España (Antena 3)
- Enfermedad rara de Menkes: Marco, el único niño del mundo con una terapia excepcional (EFE)
- La mejoría de Marco abre la puerta a otros niños con la enfermedad de Menkes (elDiario.es)
- https://www.lavozdelanzarote.com/actualidad/sociedad/juanito-enfermo-menkes-mas-longevo-mundo-recibe-tratamiento-en-lanzarote-podria-salvarle_231354_102.html

Funds spent: €45,038 *(Core´s fees, medical missions and assistance to families)*

10. Financial Accountability (Summary)

At MIa, we are committed to ensuring full transparency and responsible management and use of all financial resources. Despite operating with limited income, we have established robust financial controls to maintain the highest standards of accountability. Our financial oversight includes two layers of control: first, Belén B., an accountant specializing in the financial management of small organizations, and second, the independent firm Medina Rubio Asesores, led by an experienced economist.

In 2024, MIa raised a total of €149,950, and allocated €96,967.37 towards key initiatives. These funds supported vital areas such as the provision of treatments, travel assistance for families, and the organization of networking events. We also rely heavily on the invaluable contributions of volunteers, ensuring that the majority of our funding goes directly to supporting children with Menkes disease and their families.

Total funds raised: €45,038 *(Core´s fees, medical missions and assistance to families)*

Summary of expenses:

Activity 1: **€00.00**
Activity 2, 4 & 9: **€43,002** *(Core services and medical missions)*
Activity 3: **€7,685.97**
Activity 5: **€635.88** *(Eurordis and BlueHost)*
Activity 6: **€31,156**
Activity 7: **€ 11,180**
Activity 8: **€1,090**
Activity 9: **€2,036** *(excluding core´s fees, medical missions and assistance to families)*
Activity 10: **€181.50** -Accountant fees *(Medina Rubio asesores)*

TOTAL expenses: €96,967.37

Net income for MIa: €52,982.67

Menkes International Association Boards (2024)

The Executive Board of MIa is composed of dedicated individuals with personal connections to Menkes disease. They guide the organization's day to day operations and ensure that MIa continues to serve the families and children affected by this rare condition.

- **Aurora Mateos** *(Executive Secretary) – Menkes relative*
- **Tote Portillo** *(Secretary) – Menkes relative*
- **Vicente Mateos** *(Treasurer) – Menkes relative*
- **Denis Broun** *(Medical Director)*

Advisory Board

The Advisory Board consists of experts from various fields who provide strategic advice and direction. While some members have personal connections to Menkes, others bring external expertise to strengthen MIa's work.

- **Denis Broun** *(Pharmaceutical Adviser) – Non-Menkes relative*
- **Leonardo Cervera Navas** *(Data-Policy Adviser) – Non-Menkes relative*
- **Andrey Galaev** *(Adviser) – Menkes relative*
- **Jorge A. López** *(IT Manager) – Menkes relative*
- **Juan Mateos** *(Adviser) – Menkes relative*
- **Inma Mateos** *(Communications) - Menkes relative*
- **Irene Melo** *(Marketing) – Menkes relative*
- **Jean-Francois Pulvenis** *– Non-Menkes relative*
- **Vinay Saldanha** *(Policy Adviser) – Menkes relative*

Copperless Committee

The Copper(less) Committee is an international panel of esteemed scientists and medical professionals who specialize in copper metabolism and Menkes disease. This multidisciplinary team advises MIa on the latest research and treatment developments, contributing significantly to the progress made in treating Menkes patients.

- **Prof. Francesc Palau Martínez** *– Director, Copper(less) Committee; Director, Genetics and Molecular Medicine, Institut de Recerca Sant Joan de Déu, Barcelona, Spain*
- **Dr. Denis Broun** *– CEO, Givopax Pharmaceutical Company, Geneva, Switzerland*
- **Dr. Mercé Capdevila** *– Professor, Dept. of Chemistry, Universitat Autònoma de Barcelona, Spain*
- **Prof. Vishal Gohil** *– Associate Professor, Biochemistry & Biophysics, Texas A&M University, USA*
- **Dr. Elena Godoy** *– Specialist, Pediatric Complex Care and Palliative Care Unit, Regional University Hospital, Málaga, Spain*
- **Prof. Nina Horn** *– Retired Pharmacist and Menkes Researcher, Kennedy Center-Copenhagen University Hospital*
- **Prof. Svetlana Lutsenko** *– Professor of Physiology, Johns Hopkins University School of Medicine, Baltimore, USA*

- **Aurora Mateos** – *Leader of MIa, mother of Marco (first patient treated with Elesclomol-Copper)*
- **Prof. Victor Mangas** – *Assistant Professor, University of Valencia, Spain, Specialist in Pharmaco-kinetics and Bioequivalence*
- **Prof. Michael Petris** – *Professor of Biochemistry, University of Missouri-School of Medicine, Columbia, USA, Menkes Researcher*
- **Prof. James Sacchettini** – *Rodger J. Wolfe-Welch Foundation Chair of Science, Biochemistry & Biophysics, Texas A&M University, USA*
- **Prof. Joseph Standing** – *Professor of Pharmacometrics, UCL Institute of Child Health, London; Research Pharmacist, Great Ormond Street Hospital, London*
- **Dr. Prachi Trivedi** – *Associate Research Scientist, Gohil Lab, Texas A&M University, USA*

Menkes International Association - Aurora Mateos.