VOLUME 2

MAR - AUG 2025

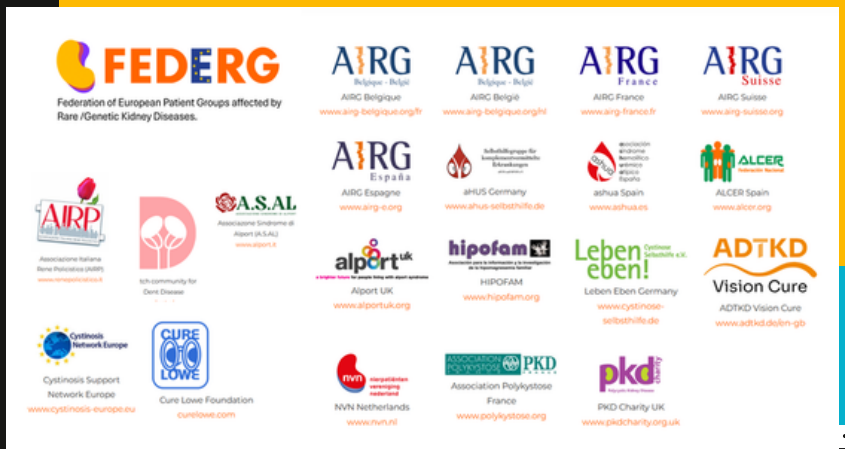
MEMBERS ACTIVITIES Newsletter

spring- summer

EXPERIENCE THE

Magic of Summer

with Our Exclusive Federg Members Activities Last News!



LET US HELP YOU CREATE SUMMER MEMORIES THAT WILL LAST A LIFETIME.

VOLUME 2
MAR – AUG 2025

FEDERG
European Patient Associations affected by Rare / Genetic Kidney Diseases.

MEMBERS ACTIVITIES
Newsletter
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Magic of Summer

with Our Exclusive Federg Members Activities Last News!

Susana Carvajal Arjona
President
FEDERG Federation of European Patient Associations affected by a rare and genetic kidney diseases

July 2025

Summer is here, bringing longer days and new adventures! It's the perfect moment to discover what our Federg Membership Associations have achieved in recent months and what inspiring plans they have for after the summer: supporting patient communities and joining forces for campaigns that highlight their needs and help improve their quality of life.

Patient associations never stop — they give their very best to reach families living with rare kidney diseases, making sure they are never alone and always have the support they need.

Let's kick off this newsletter, volume 2 by celebrating our partners' inspiring activities!

Raising awareness and advocating for better care, to providing vital support and creating spaces where no one feels alone — our community never stops. Together, we turn hope into action and support into strength. Let's keep moving forward, united by our commitment to improve the lives of everyone living with a rare kidney condition.

- The 20th Anniversary of AIRP
- Italian Polycystic Kidney Disease Associaton ETS.
- Special Dinner and amazing patient participation in within the framework of ERA Congress. MPGN and aHUS.
- 5th National Alport 2025 Congress in Turin, ASAL.
- Pediatric transplant, living donor, a chance at life, Hipofam Association.

New FEDERG members:

- Lowe Foundation by Sébastian Honoré, Giving voice to Lowe syndrome patients in Denmark.
- Anne Marie Pickaert, Renaloo, France. Representative of patients with various kidney diseases.
- ASHUA Celebrates Its XII Conference in Seville.
- AIRG-E Spain Brings Patients Together: A Double Event in Barcelona and Madrid.

The 20th Anniversary of AIRP Italian Polycystic Kidney Disease Associaton ETS

AIRP is thrilled to celebrate the twentieth anniversary of our partnership with the Conference:
"20 Years of Unwavering Commitment in the Fight Against Polycystic Kidney Disease — Standing Shoulder to Shoulder with Patients Every Step of the Way."

Where and when?
Regione Lombardia - Auditorium Testori
Piazza Lombardia 1 – Milan
September 20, 2025
8:30 AM - 6:00 PM.

The main goal was to increase the visibility of the disease, raise public awareness, and improve the quality of life for patients through accurate information, psychological support, and educational iniciatives.

The Italian Polycystic Kidney Disease Associaton ETS (AIRP) was founded twenty years ago with the aim of supporing individuals affected by this rare genetic disease and promoting scientific research to improve the quality of life of patients.
The history of the Association is marked by commitment, solidarity, and significant progress in the field of renal health.

Foundation and Early Years

The Association was founded in 2005 by a group of patients, family members, and doctors, united by the determination to create a support network for those living with polycystic kidney disease. Initially, the Associa0on focused its efforts on raising awareness of this condition and creating a forum to facilitate the exchange of experiences among patients.

Unfortunately, polycystic kidney disease is a rare condition, and patients often face the diagnostic and therapeutic journey in isolation.

Growth and Awareness

Over the years, AIRP has significantly expanded its activities, dedicating itself to organizing awareness, events, conferences, and informational meetings. These events had a notable impact, enabling the Association to reach a wider audience and inform both patients and healthcare professionals about the symptoms, complications, and available treatment options for the disease.
The awareness campaign also involved collaborations with other associations and institutions, creating a united front in the fight against the disease.
A crucial aspect of the Association's work has been its commitment to supporting scientific research, with the goal of discovering more effective therapies and, ideally, a definitive cure.
Starting in 2008, AIRP also began holding annual meetings to update members on the latest scientific advancements, ongoing treatments, and future prospects.

Research and Innovation

In 2010, AIRP launched important collaborations with universities and research centers to promote scientific studies on PKD. Thanks to the funds raised through the 5x1000 program, numerous research projects in partnership with experts and doctors were supported, aimed at exploring new therapeutic possibilities. The scientific community recognized the Association's contribution as an opportunity to accelerate the understanding of the disease and available treatment options.
Moreover, the Association carried out awareness projects through social media, increasing the visibility of the disease and its implications. Communication became a key element, enabling many patients to connect with the reality of the Association and with others who share the same condition.

Support for Patients

Another cornerstone of AIRP has been direct support for patients and their families. The Associatioon launched a counseling service to provide useful information regarding treatments, patient rights, and access to innovative therapies. A support group was created where patients can share experiences and find comfort within the community.

International Collaboration

In recent years, AIRP has expanded its activities internationally, establishing collaborations with patient associations and research institutions across Europe and beyond. This synergy has facilitated the exchange of knowledge, experiences, and best practices, contributing to a global understanding of the disease and its challenges.

One of the most significant recent developments has been the strengthening of international collaborations, enabling AIRP to play an active role in global research networks such as PKD International, FEDERG, ERKNet, and EURORDIS. These important and powerful alliances not only keep us informed about the latest scientific discoveries but also provide a platform for us to share our best practices, support others, and contribute to common goals alongside other patient associations.

By working together, we help shape the advocacy landscape and drive meaningful change for those affected by genetic kidney diseases. Through collaboration, we strive for a future where patients have a stronger voice, better care, and improved treatment options—bringing to life one of our favorite mottos: "Insieme si può" (Together, we can achieve it!).

AIRP, Future and New Challenges

Today, the Italian Polycystic Kidney Disease Association continues to advocate for patients' rights, working tirelessly to improve the quality of life for those affected by this disease.
As of 2025, AIRP can boast a history rich in successes. Over these 20 years, it has supported thousands of people, made significant contributions to scientific research, and raised public awareness about polycystic kidney disease. The Association has also achieved significant milestones in promoting laws that protect patients' rights and in improving their quality of life.
In summary, AIRP's twenty years represent a concrete example of how solidarity, research, and commitment can transform the lives of many people. Looking toward the future, the Association continues its mission with passion and determination, fully aware of the importance of each individual life affected by the condition.
Looking ahead, the Italian Polycystic Kidney Disease Association has chosen to continue its work, placing an increasing focus on therapeutic innovations and the creation of a stronger, more united community. Future objectives include new awareness campaigns, further research funding, and more intense interactions with health institutions to improve patient care.
In these twenty years, AIRP has become a beacon for all those living with polycystic kidney disease, offering support, hope, and resources to face the daily challenges. Its story is a testimony of commitment, determination, and passion in improving the lives of those affected by this condition.

January 2025
Luisa Sternfeld Pavia
President
AIRP • Associazione Italiana Rene Policistico
luisa.sternfeld.airp@renepolicistico.it
www.renepolicistico.it

Special Dinner and amazing patient participation in within the framework of ERA Congress. MPGN and aHUS

Selbsthilfe für seltene komplementvermittelte Erkrankungen e.V.
MPGN und aHUS

Rigth to the left 5th personne Christiane Mockenhaupt (aHUS Germany president)

Alongside the recent ERA Congress in Vienna, patient representatives were invited too participate — marking an important step forward in collaboration between medical experts and those directly affected by rare kidney diseases. Christiane Mockenhaupt, president of Selbsthilfe für seltene Komplementvermittelte Erkrankungen e.V., took part to share the patient perspective on atypical Hemolytic Uremic Syndrome (aHUS).

aHUS is a very rare, serious kidney disease caused by uncontrolled activation of the complement system — a part of our immune system. This leads to the destruction of red blood cells and damage to the small blood vessels in the kidneys and other organs. Without timely diagnosis and treatment, aHUS can cause kidney failure and other severe complications.

"I had the chance to tell our story and highlight the emotional side of living with a rare disease like aHUS, Christiane said. ""Our journey began 13 years ago, when there was almost no information about aHUS online.

We felt completely alone and were fortunate to find a community of kidney disease parents at our university hospital who supported us just by listening. That inspired us to build something bigger, so that no one would ever feel as isolated as we did."
A year later, I founded our own patient support group. Since 2014, we have organized an aHUs Annual Patient Day to share knowledge, raise awareness, and connect patients and families who often feel invisible.

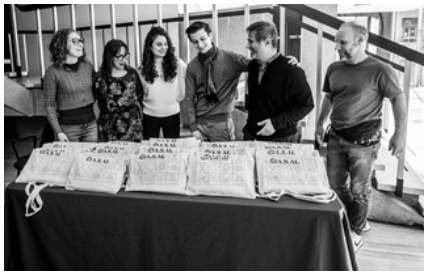
"This November, we will host our second International aHUS and C3G/IC-PGN Patient Day in Heidelberg. Everyone is welcome — patients, families, caregivers, and healthcare professionals," Christiane shared. "We invite doctors to pass this information to their patients so they can expand their support network and exchange experiences."
For people living with rare diseases like aHUS, community is crucial. By coming together, patients and families can share practical advice, emotional support, and, importantly, work together to improve diagnosis, treatment, and care. Sharing our stories helps raise awareness among doctors and policymakers, pushing for earlier diagnosis, better therapies, and more research into rare kidney diseases.
"Alone, we may feel powerless — but together, we can make our voices heard and create real change," Christiane said. "We warmly invite everyone to join us, learn from each other, and help ensure that no one facing a rare kidney disease ever feels alone again."

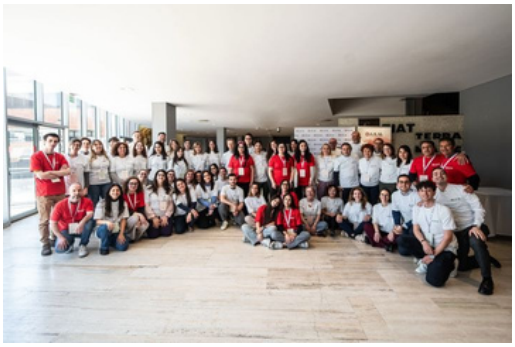
June 2025
Christiane Mockenhaupt
President
Selbsthilfe für seltene Komplementvermittelte Erkrankungen e.V
christiane.mockenhaupt@ahus-selbsthilfe.de;

5th National Alport 2025 Congress in Turin, ASAL

A.S.AL
ASSOCIAZIONE SINDROME DI ALPORT
Associazone Sindrome di Alport
(A.S.AL)

The Italian Alport Syndrome Association has organized this year's National Congress with the aim of offering a comprehensive update to healthcare professionals in various related fields (adult and pediatric nephrologists, geneticists, pathologists, otolaryngologists, ophthalmologists, psychologists, nutritionists) as well as to patients, sharing the entire event together.



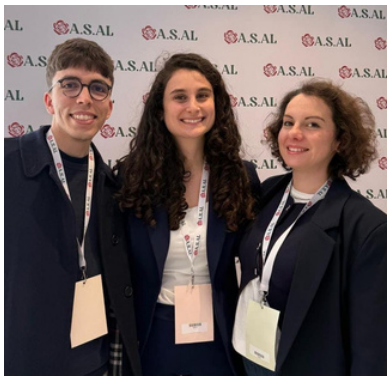


Rigth to the left Margherita Venturini, patuient reprersentative, Daniela Lai, Asal president, Angela María Ferrante, Asal vice president.

5th National Conference on Alport Syndrome: Challenges and Perspectives in Clinical Practice April 11-12, 2025 – Turin, National Automobile Museum.

Alport Syndrome is currently one of the most prominent diseases in the international nephrological scientific community, both in terms of clinical research and basic research. The structural alteration of the glomerulus underlying this disease is a biochemical model that is as complex as it is intriguing for understanding the functioning of the filtration unit and for the development of new drugs.
The Italian Alport Syndrome Association has organized this year's National Congress with the aim of offering a comprehensive update to healthcare professionals in various related fields (adult and pediatric nephrologists, geneticists, pathologists, otolaryngologists, ophthalmologists, psychologists, nutritionists) as well as to patients, sharing the entire event together.

The chosen venue was Turin, a city that has always combined art, science, and innovation, and is a well-established host of major international events.
The congress spread over two days, focusing on the state of the art in genetics and basic research, the clinical and therapeutic aspects of the disease, and the Association's projects where doctors and patients meet for the continuous improvement of the quality of care for Alport Syndrome patients and their families.



Right to the left: Elia Galli, Margherita Venturini, Giulia Nutile (Patient representatives for Rete_Alport project)

A.S.AL (Alport Syndrome Association ODV) is a non-profit organization founded in Sassari, Sardinia, and has since expanded its activities across Italy and internationally. Since 2011, A.S.AL has been working to bring together healthcare professionals, promote and support initiatives aimed at improving information, care, and assistance for Alport patients and their families.

Alport Syndrome is a genetically determined disorder caused by pathogenic variants in the COL4A3, COL4A4, and COL4A5 genes, which affect the structure of type IV collagen in the basement membrane. Since the COL4A5 gene is located on the X chromosome and COL4A3 and COL4A4 are located on chromosome 2, three primary modes of inheritance have been identified: X-linked, autosomal recessive (AR), and autosomal dominant (AD).
These pathogenic variants compromise the normal structure and function of the basement membranes in the kidney, inner ear (cochlea), and eye, leading to progressive kidney damage, hearing loss, and ocular abnormalities in many affected individuals. The clinical manifestations of Alport Syndrome develop over time and are often accompanied by a significant psychological and financial impact on patients and their families.
As a rare multisystemic genetic disease, Alport Syndrome presents the typical challenges associated with rare conditions: difficulty in obtaining a timely diagnosis, lack of dedicated reference centers, complex clinical management, and significant social and psychological repercussions for both patients and their families.

The idea to create the association stemmed from a desire to contribute to the understanding, study, and scientific research of Alport Syndrome. Building a network between healthcare professionals and families has been the Association's core mission from the start. Over the years, A.S.AL ODV has organized various national conferences to support the training of healthcare providers, facilitate dialogue with patients, and discuss diagnostic and therapeutic pathways.
Providing support and staying close to families has become a cornerstone of A.S.AL's activities. Living with Alport Syndrome can have a profound psychological impact—not only on those diagnosed, but also on their families. The fact that multiple members of the same family may be affected can intensify the emotional burden. Key factors impacting the quality of life for young patients include, for example, difficulties in using hearing aids, adherence to pharmacological treatments, and the need to follow a specific diet.
A.S.AL has launched several initiatives to offer emotional support and a space for dialogue for young Alport patients and their parents, guided by professional facilitators. One of these initiatives is the "Alport - Never Alone" project, which aims to improve the quality of life for individuals with Alport Syndrome, both young and adult. The project offers a safe space for sharing experiences and developing shared, alternative solutions to everyday challenges. It also promotes relationship skills, resilience, and life skills that help individuals cope with a rare genetic condition.
Another major goal of A.S.AL is to ensure that all patients with Alport Syndrome have access to multidisciplinary and equitable care throughout Italy. Due to the large number of pathogenic variants, diverse inheritance patterns, and varying severity of symptoms, the clinical spectrum of Alport Syndrome is extremely broad. As a result, patients require care from multiple specialists throughout their lives. A multidisciplinary and easily accessible approach is essential for properly meeting the needs of individuals living with a chronic condition like Alport Syndrome.

To address this, the Association launched the “Alport Network” project, aimed at creating a network of reference centers specialized in diagnosing and managing Alport Syndrome. The project ensures that patients and families receive the necessary care and assistance. Through this initiative, A.S.AL also acts as a bridge between hospitals and patients, as well as a resource for general practitioners (GPs) and pediatricians, providing them with tools to properly guide patients with suspected kidney disease toward appropriate diagnosis and care.

A significant milestone was the 5th National Alport 2025 Congress, held in Turin on April 11–12, which saw wide participation from doctors and patients, featuring international guests and marking both a culmination and a new beginning for future scientific and support initiatives.

For more details about A.S.AL’s projects, activities, and upcoming events, we invite you to visit the Association’s official website: www.alport.it

April 2025
Giulia Nutile
Patient representative
ASAL

hipofam
Asociación para la información y la investigación de la hipomagnesemia familiar
HIPOFAM

Pediatric transplant, living donor, a chance at life, Hipofam Association.

When a rare kidney disease causes kidney failure, a transplant brings hope and a new chance at life. This pre-summer, the Association for Research and Information on Familial Hypomagnesemia — Hipofam — is shining a light on the life-changing option of kidney transplantation. Pediatric kidney transplants are not just a medical procedure; they’re a lifelong journey of love, resilience, and family togetherness.

This year, at the Hipofam patient association, we stood beside brave adolescents and their families as they faced one of life’s greatest trials: kidney failure. Some families chose the courageous path of a living donor transplant — a gift of life given from one body to another. This therapy is far more than a medical procedure; it’s an act of faith, sacrifice, and unyielding love.

To choose this path means to watch a kidney fail and witness the young spirit it sustains begin to fade — only to see hope reignite when that organ is reborn in another body. It means facing fears head-on, placing trust in science and in each other, and daring to believe in life again.

But this second chance comes with challenges. It demands strict routines, avoid solar radiation that can cause skin lesions and cancer and social restrictions to prevent infections and contagions and powerful medications that protect the new organ. Medications often bring side effects that can be as daunting as the disease itself — diabetes, swelling, tremors, mineral imbalances, blood pressure, acne, alopecia, hyperglycemia and diabetes and surgical complications that require expert support, providing frequent monitoring that will help life gradually return to normal.

For a family this means challenges before and after transplant, remain firm, calm and positive to provide the best support to two people in a family. It means two family members entering the operating room together: one giving, one receiving, both forever changed. It's a family challenge, the caregiver's role is essential, it's a team effort.

In these moments, emotional support is not just important — it is vital. Families must prepare not only before the transplant but long after it, as they navigate this new life together. They must learn to care for this precious gift, this kidney that now beats with two hearts’ hope. They must stay vigilant, because a transplant is not a cure — it is a commitment to protect life, every single day.

And yet, with care, with love, with guidance and community, the fear softens. The side effects fade into the background. And what remains is a young person who can dream again, run again, live again — differently, yes, but fully. A future that depends not only on medicine, but on the unwavering love and responsibility of a family who chose life, together.

spring- summer

FEDERG
European Patient Associations affected by Rare / Genetic Kidney Diseases.

Mar- Aug 2025

Rigth to the left Aram Rodríguez López (Young Patient with HFHN, organ recipient, Miriam López Pérez,live donor)

Rigth to the left Antoni Cabrera C. (Young Patient with HFHN , organ recipient , Antonio Cabrera (live donor, hipofam president)

A Piece of Life, from Father to Son

There are actions that transcend words, decisions that are engraved in the depths of the soul. Donating a kidney is giving life, but when that gift comes from a father/mother to his son, it becomes a pure testament of unconditional love. This isn't just a medical gesture: it's a silent promise that says, "I'll do anything to see you live, to see you smile, to give you a new chance." It's the heart of a father/mother beating strongly in two bodies, united not only by blood, but now also by hope. In this story, there are no invisible heroes, because a father's love shines with a light that no one can ignore. A kidney changes a destiny, but the act of giving it changes the very meaning of the word "life." May this renewed bond be a symbol of strength, of the future, and of eternal gratitude. Because when a father gives a part of himself to save his son, the miracle has already begun.

June 2025
Susana Carvajal Arjona
President
Federg and co-founder hipofam
hipofam.org

Bringing Visibility and Action: Cure Lowe Foundation Joins FEDERG

Cure Lowe Foundation was founded by Sébastian Honoré, an economist and father to Walther, a young boy living with Lowe syndrome — a rare genetic condition that affects the kidneys, brain, and eyes from birth. Based in Copenhagen, the foundation emerged from a deeply personal need to accelerate research and find better treatments, not only for Walther but for all children and families facing this devastating diagnosis.

With a background in public finance and a growing European network of scientists, clinicians, and families, Sébastian now leads the foundation's mission to develop the first gene therapy for Lowe syndrome, with an initial focus on protecting kidney function. Cure Lowe brings together professional expertise and lived experience to advocate for earlier diagnosis, equitable care, and, crucially, stronger patient involvement in research and decision-making.

As a community representing patients who have long been underrepresented, Cure Lowe believes it is vital to remain active, visible, and collaborative. The foundation is committed to raising awareness and driving progress — not just for Lowe syndrome, but for the broader rare kidney disease community.

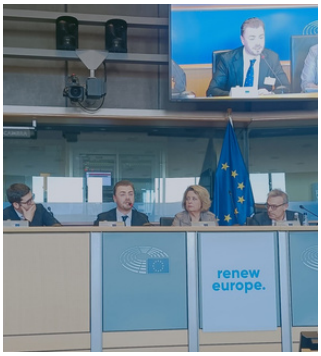
Cure Lowe is proud to join FEDERG and looks forward to contributing actively to our shared efforts to improve the lives of all patients living with rare kidney diseases across Europe

From Personal Fight to Collective Action: Cure Lowe Stands Up for Rare Kidney Diseases in More than you can imagine. Conference at the European Parliament, March 2025.

Sebastian Honoré, founder of the Cure Lowe Foundation, shared a moving and deeply personal story about his son Walther's fight against Lowe Syndrome — an ultra-rare genetic disorder for which no treatments currently exist. He emphasized that this absence of treatment is not because science lacks the answers, but because economic obstacles stand in the way of translating research into real therapies.

"We are standing on the threshold of a major breakthrough," he explained, pointing to the significant scientific progress made in understanding the underlying biology of Lowe Syndrome and developing potential therapeutic approaches, including gene therapy. Yet, he cautioned that the enormous financial risk and limited commercial incentives for ultra-rare conditions often deter investors and pharmaceutical companies from stepping in, leaving families and small patient-led foundations to shoulder the burden alone.

This, he stressed, is a clear example of the so-called "orphan drug paradox": despite the existence of cutting-edge science and dedicated researchers, lifesaving treatments for ultra-rare diseases remain out of reach simply because the patient population is too small to attract the investment needed to bring promising ideas to life. Honoré called for greater collaboration, innovative funding models, and collective action to break through these economic barriers — so that scientific progress can finally translate into real hope and tangible solutions for families who have waited too long.



June 2025
Sebastian Honoré
President
Cure lowe Foundation
curelowe.com

Renaloo: Strengthening European Collaboration to Make Kidney Disease a Public Health Priority

Renaloo
La voix des malades du rein

Renaloo is a leading French patient association dedicated to supporting people affected by chronic kidney disease (CKD). Representing a vibrant community of over 8,000 patients and caregivers, Renaloo brings together individuals living with a wide range of kidney conditions, including many who face rare and inherited kidney diseases.

At Renaloo, we believe that collaboration and solidarity are essential to driving real change. We share common values with Federg, the European Federation of Patients with Rare and Genetic Kidney Diseases, and we are deeply committed to working together to build a stronger, more unified voice for kidney patients across Europe. By combining our experience, networks, and advocacy efforts, we can help ensure that meaningful actions — especially in prevention, early detection, and equitable care — are implemented consistently throughout the European Union.

"Raising our voice within European institutions is a vital step toward improving patient care and securing greater recognition of kidney disease as a major public health challenge," says Anne-Pierre Pickaert, Renaloo's new representative at Federg.

Anne-Pierre Pickaert brings to this mission more than 20 years of expertise in public health, patient access, and healthcare policy. Her extensive experience spans the charity sector, patient organizations, government agencies, consulting, and the pharmaceutical industry — a background that gives her a comprehensive understanding of how to bridge stakeholders and advance impactful solutions.

As Renaloo's voice within Federg, Anne-Pierre sees this new partnership as a powerful way to amplify the perspectives of patients with rare and genetic kidney diseases. "Joining forces allows us to pool our knowledge, share best practices, and strengthen our collective ability to advocate for everyone affected — regardless of where they live or which rare condition they face," she explains.

Together, Renaloo and Federg are determined to tackle challenges that patients continue to face — from late diagnoses and limited treatment options to gaps in social and psychological support. By speaking up at the European level, they aim to secure better research funding, promote harmonized standards of care, and push for policies that address kidney disease prevention and early intervention as urgent priorities.

June 2025
Anne-Pierrem Pickaert
Renaloo
renaloo.com

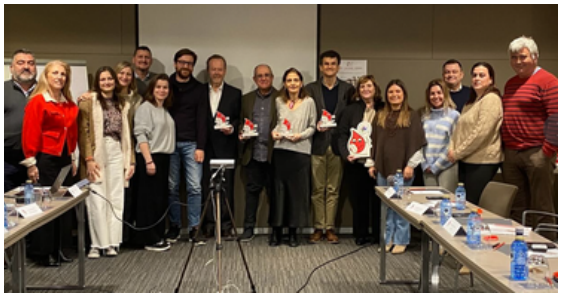
spring- summer

FEDERG
European Patient Associations affected by Rare / Genetic Kidney Diseases.

March- August 2025

ASHUA Celebrates Its XII Conference in Seville to Inform and Educate Patients and Families

Seville, February 15, 2025. The Atypical Hemolytic Uremic Syndrome Association (ASHUA) has held its XII Conference in Seville with the aim of continuing to inform and educate patients and their families. The event was supported by nephrologists from the Virgen del Rocío University Hospital, reaffirming the importance of knowledge and research in improving the quality of life for those affected by this rare disease.



According to the association: "Informing and educating patients is key. Only by empowering them can we save lives and demand greater investment in research from the Public Administration. Despite progress, there are still delays in diagnoses and patients without access to adequate treatment, putting their lives at risk."

Challenge in the Field of Rare Diseases

Atypical Hemolytic Uremic Syndrome (aHUS) is one of the 7,000 identified rare diseases, of which only 10% have an effective treatment. Since 2011, aHUS patients have had access to life-saving treatments, but ASHUA insists that without research and early diagnosis, the disease will continue to cause irreversible damage.

"In 2024, thirteen years after the arrival of the first medication for aHUS, there are still patients losing kidney function due to the lack of training among some doctors or budgetary restrictions from the public administration. How much is a person's life worth?" the association questions.

Expert Lectures: Advances and Challenges

The XII Conference featured participation from renowned specialists. Dr. Francisco de la Cerda addressed the diagnosis of aHUS in childhood, assuring that Spanish hospitals are equipped to detect it quickly, although he emphasizes the need for continued training for healthcare professionals.

Dr. Virginia Cabello discussed the relationship between aHUS and motherhood, noting that while pregnancy was previously contraindicated in these cases, recent studies have shown it is possible with proper precautions and monitoring.

Another key moment of the conference was the intervention of Pablo Beca, president of ALCER Giralda and ALCER Huelva, who highlighted the importance of organ donation. "Donating life costs nothing," he emphasized, reminding that Spain continues to be a global leader in donations.

Finally, Dr. José Luis Rocha, head of the Nephrology Department at Virgen del Rocío Hospital, emphasized the advances in Spanish healthcare and the high qualification of its professionals, an aspect that ASHUA also acknowledges and supports.

spring- summer

FEDERG
European Patient Associations affected by Rare / Genetic Kidney Diseases.

Mar- Aug 2025

The Importance of Knowledge and Education

ASHUA remains committed to providing rigorous information so that patients can lead a normal life as long as they receive the appropriate treatment. “If any of these factors fail, patients' lives are at risk,” warns the organization.
Fortunately, more and more healthcare professionals are collaborating with ASHUA to disseminate medical advancements. The association highlights their selfless work, as many of these specialists dedicate their free time to educating patients without receiving support from the Public Administration.

Tireless Work in Defense of Patients

ASHUA has worked intensively to position the association and aHUS patients at a prominent place within the healthcare, political, and social spheres. Its president, Francisco Monfort, shared his personal experience, recalling that the association was founded in 2012 after a five-year struggle for his son’s life. “Doctors gave us a bleak prognosis in 2006, but research changed our son's destiny. It was our duty to share this experience and help other patients.”
Since 2015, ASHUA has been recognized as a Public Utility Entity and is part of the National ALCER Federation. Among its initiatives is the creation of a QR bracelet so that patients can be quickly identified in case of a medical emergency. Additionally, it has launched the ASHUA Observatory, a tool that evaluates medical care, patients’ quality of life, and identifies urgent needs in aHUS management.
At the international level, ASHUA has expanded its work. Since 2014, it has been a founding member of the European Federation of Genetic Renal Diseases (FEDERG) and participates in the European Reference Network for Rare Kidney Diseases (ERKNET), representing the patients' voice.

Research: Key to the Future

Despite progress, much work remains. Research continues to be fundamental to better understand aHUS and expand genetic knowledge, as currently only 50% of the genetic defects associated with the disease have been identified.
In Spain, aHUS affects approximately 1,200 people and is one of the few rare diseases with an effective treatment. According to data from the ASHUA Observatory, 70% of cases appear before the age of 15, and 66% affect women, many of them after pregnancy. Only 5% of diagnoses correspond to people over 50 years old. Furthermore, patients demand more investment in research and access to updated information.
ASHUA’s XII Conference has reaffirmed the importance of joint efforts among patients, doctors, and researchers to continue advancing in the understanding and treatment of aHUS. The association will continue its fight to ensure that no patient is left untreated and that research remains a priority on the healthcare agenda.

March 2025
Mireya Carratalá
ASHUA
ashua.es

AIRG España
AIRG Espagne

AIRG-E Spain Brings Patients Together: A Double Event in Barcelona and Madrid to Advance Knowledge and Support

This year, the Association for Information and Research on Genetic Kidney Diseases (AIRG-E Spain) is going a step further in its mission to support, inform, and empower patients and families affected by rare genetic kidney disorders. For the first time, the association will host a hybrid double event in two major Spanish cities — Barcelona and Madrid — combining in-person and online participation to make the sessions as accessible and inclusive as possible.
The events are designed as a unique opportunity for patients, families, and healthcare professionals to come together, share experiences, and learn about the latest developments in genetic kidney diseases. The program aims to address both medical advances and the real-life challenges that patients face every day.

The Barcelona session, taking place on October 4, will focus on a broad range of topics to help patients better understand the genetic basis of kidney diseases and how this knowledge translates into better care. Attendees will hear about the fundamentals of genetics in nephrology, new clinical guidelines for rare conditions such as Alport syndrome, and practical insights into living with adult cystinosis. Other talks will cover tubular disorders, amyloidosis, and surgical aspects like nephrectomy. The event will also include an important discussion on the social and legal aspects of living with a disability, offering guidance on patients' rights and support resources.

The Madrid conference, planned for January 17 at a prominent cultural venue, will continue this patient-centered approach with sessions on cutting-edge research and daily living strategies. Topics will include the emerging role of polygenic risk scores in predicting disease risk, specific challenges faced by women with polycystic kidney disease, and an in-depth look at C3 glomerulopathy, a rare complement-mediated disorder. Participants will also learn about the latest findings on how mitochondrial diseases can affect kidney function — an area of research that recently won special recognition for its innovative contribution. The program will round out with practical advice on nutrition, highlighting how dietary choices can support better outcomes for patients with chronic kidney conditions.

By bringing together specialists, researchers, and patient advocates, AIRG-E Spain hopes to foster dialogue and build a stronger sense of community. The association's goal is not only to share information but also to ensure that patients and their families feel supported, heard, and better equipped to manage the challenges of living with a genetic kidney disease.
For more information and registration details, visit AIRG-E Spain's website and follow their channels to stay updated. Together, we can make a difference — one connection at a time.

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Marta Roger
AIRG-E
president