
# 6th INTERNATIONAL CONFERENCE ON RARE DISEASES

# POST-EVENT PRESS RELEASE

**Athens April 6th:** With the participation of 300 delegates from more than 31 countries, the 6th International Conference on Rare Diseases was successfully completed, marking another meaningful milestone for the global rare disease community.

The two-day conference under the **theme "Building a Brighter Future Together: Converting Rare Disease Challenges into National and International Opportunities",** held under the auspices of the Ministry of Health, brought together stakeholders for meaningful dialogue, collaboration, and forward-looking exchange, fostering a strong sense of shared purpose and collective responsibility. Across 10 dynamic sessions, it underscored the critical role of international cooperation, innovation, and policymaking in improving access to care for people living with rare diseases.

## Day 1: "Global Landscape & WHA Resolution: Setting the Scene for a Coordinated European Vision and Regulatory Evolution in Rare Diseases"

***The first day explored the global landscape of rare diseases, including international collaborations, regulatory science and the future of access to innovative therapies. It laid the foundations for a more coordinated European approach to Rare Diseases, emphasizing international collaborations (ERDERA, ERNs), the acceleration of Innovation through regulatory developments, the transition from Research to Treatment, and the need for fairer and more prepared access and HTA system, with an active role for Patients and the utilization of data***.

Landmark collaborations for rare diseases, as well as new policies being designed in Europe and globally, were presented by speakers **Daria Julkowska**, Scientific coordinator of the European Rare Diseases Research Alliance (ERDERA)/Assistant Director of the Thematic Institute for Genetics, Genomics & Bioinformatics (IT GGB) at INSERM, **Maurizio Scarpa**, Coordinator at the European Reference Network For Rare Hereditary Metabolic Diseases METABERN, **Holm Graessner**, Managing Director, Centre for Rare Diseases, **Ines Hernando**, ERN and Healthcare Director EURORDIS, and **Monica Drum**, Global Programmes Senior Manager, Rare Diseases International, in the first panel of the 6th Conference on Rare Diseases. The session was moderated by **Ana Rath**, Orphanet Director & OD4RD2 coordinator, commissioner for Rare Diseases International Lancet Commission on Rare Diseases.

International and European collaborations are emerging as a key driver of progress for rare diseases, shaping a new, more coordinated policy landscape. The new European plan, combined with initiatives in the US and the WHO, strengthens collective action on a global level. The role of European Reference Networks (ERNs) is decisive, coordinating


hundreds of specialized centers and promoting research into diagnoses and innovative treatments.

The **ERDERA alliance**, with broad international participation and significant funding, aims to accelerate diagnosis and treatment, as well as improve patients' quality of life. At the same time, emphasis is placed on strengthening national policies and linking them with the international ecosystem. Particular importance is also attached to supporting undiagnosed patients and enhancing the "visibility" of rare diseases. The need for data collection and utilization by all involved stakeholders for more effective interventions was also emphasized. International organizations promote tools and directions, especially for countries with limited resources, aiming to reduce inequalities in access and care.

**Regulatory Science and Therapeutic Innovation**

Promising developments regarding shorter approval timelines for orphan drugs by the European Medicines Agency (EMA) under the new European pharmaceutical legislation, as well as points of concern, were discussed by representatives of patients, industry, and competent EU regulatory bodies in the panel titled **"Regulatory Science & Innovation Pathways."** The session was moderated by **Dimitrios Athanasiou**, VP Rare Diseases Greece, EMA PCWP Member, GPA WDO, BoD, and **Victor Maertens**, Government Affairs Director at EUCOPE.

Specifically, **Kaja Kantorska**, Policy Officer, European Commission, **Violeta Stoyanova-Beninska**, Senior Scientific Specialist at Human Division at European Medicines Agency, Ex-Chair of COMP of EMA, Chair of Regulatory Science Committee of International Rare Disease Research Consortium (IRDIRC), **Claudia Louati**, Head of Policy, European Patients' Forum (EPF), **Dominika Misztela**, PhD Director, Global Regulatory Science, Policy and Intelligence - Europe, Global Regulatory Affairs, Research and Development (GRA R&D), CSL, and **Kavita Patel**, Managing Director, Roche Hellas SA in Greece & Cyprus, focused on the new European pharmaceutical legislation. This legislation is expected to significantly accelerate orphan drug approvals by the EMA, emphasizing unmet medical needs and patient-centered policies. Of particular interest are the "regulatory sandboxes," which strengthen cooperation between agencies and accelerate innovation. Meanwhile, increasing patient participation in decision-making is a positive development, although concerns were expressed regarding the reduction of experts.

Weaknesses of the framework were also highlighted, such as the limited coverage of all rare diseases and the non-differentiated reward of innovation. Industry representatives stressed the need for early dialogue with authorities and a stable regulatory environment. Finally, implementation challenges at the national level were noted, with Greece as a characteristic example, where reimbursement procedures may delay access, also affecting Europe's competitiveness.


In the 3rd Session **" Therapeutic Development & Evidence Generation"** featuring **Christina Kyriakopoulou**, PhD, Policy Officer, Health Research Programmes - Scientific advice: Rare diseases, Health data, AI in health research, **Giada Farinelli**, Regulatory Affairs Manager at Fondazione Telethon, **Rose Maase**, Therapeutic Operations Lead (Benelux) at Orchard Therapeutics, **Sarah Snedeker**, Senior Director of Pipeline Strategy, Chiesi, and moderated by **Stefano Benvenutti**, Public Affairs manager, Fondazione Telethon, platform technologies and new ATMP production tools were highlighted as catalysts for accelerating treatment development for rare diseases. Platforms allow the use of a common "backbone," facilitating and accelerating approvals for multiple indications. Particular emphasis was placed on decentralized production (bedside manufacturing), which allows for faster and safer administration of personalized therapies. It was stressed that this model significantly reduces the costs and risks of transporting biological material.

At the same time, ultra-rare diseases remain a challenge, as the traditional commercial model is not sustainable. In this context, the increased role of the academic community and non-profit organizations in treatment development was highlighted. The European Union is promoting new funding tools and regulatory support to bridge the gap from research to approval. Finally, the need to strengthen regulatory readiness was noted through initiatives such as the ATMP Spearhead, for better coordination between national authorities and European organizations.

**The Biotech Act and Health System Preparedness**

The emerging European policy framework aimed at strengthening biotechnology competitiveness in Europe, accelerating innovation, and improving patient access to advanced therapies was discussed by **Jean-Philippe Plançon**, Chairman AFNP, **Fabio D'Atri**, Policy Officer European Commission, **John Coughlan**, Head of International Government Affairs and Public Policy Regeneron, **Marc van Voorst**, Head of Public Affairs Forbion, and **Calie Sharman**, General Manager Ipsen Greece, Cyprus, Israel & Malta, in **Session 4: "Biotech Act."**

The session focused on critical issues for the European ecosystem: equitable patient access to treatments, shortening timelines for Clinical Trials (including Advanced Therapy Medicinal Products - ATMPs), and what the Biotech Act means for Orphan Medicinal Products (OMPs) and the EU's attractiveness. Concurrently, a discussion opened regarding a potential "Strategic Project on Rare" and the need to cover the investment gap. The moderators were **Victor Maertens**, Government Affairs Director EUCOPE - European Confederation of Pharmaceutical Entrepreneurs, and **Dimitrios Athanasiou**, MBA, Vice President RARE DISEASES GREECE.

The **5th session, "Access, HTA & Health-System Preparedness,"** moderated by **Valentina Strammiello**, Director of Programmes European Patients' Forum, and


featuring panelists **Julie SPONY**, Project Officer HTA European Commission, **Anja Schiel**, Special Adviser NoMA, and **Thomas Grub**, Director Global Pricing and Market Access Medac, focused on the future of European Health Technology Assessment (HTA). This included the transition from EUnetHTA 21 to the implementation of Joint Clinical Assessments (JCA), as well as the readiness level of member states for their integration into national decisions. Additionally, the role of data and structured patient participation was highlighted, along with initiatives such as the WHO Novel Medicines Platform.

The first day concluded with closing remarks from **Pr. Mohamed Hassany**, Assistant Minister of Health for Projects and Public Health Initiatives, Egypt, who, among other things, mentioned Egypt's commitment to early diagnosis through newborn screening for 19 disorders within 72 hours of birth. He also stressed the importance of the Rare Diseases Fund, which ensures equitable access to treatments based on clinical need rather than financial capacity.

His remarks reflected a broader consensus that emerged throughout the day: rare diseases are increasingly being recognized as a global public health priority. Yet, despite this momentum, major gaps persist in translating innovation into timely and equitable patient access. Discussions repeatedly pointed to the need for stronger alignment between regulatory frameworks, reimbursement systems and national implementation, as well as enhanced data sharing and cross-border collaboration.

## Day 2: Greece in the European Rare Disease Ecosystem: Rare Diseases as a National Priority

***The second day of the conference focused on the translation of European strategies into national policies and practical implementation, highlighting Greece's role within the European rare disease ecosystem. Emphasis was placed on strengthening research capacity, advancing genetic care strategies, and enhancing digital health infrastructure, including patient registries and data systems, as key pillars for improving diagnosis, care, and access for people living with rare diseases.***

The proceedings of the **6th International Conference on Rare Diseases** opened with an introductory speech by the Minister of Health, **Adonis Georgiadis**, who once again expressed his steadfast support for Rare Diseases Greece (RDG), emphasizing that "the office is always open for resolving issues."

More than 500,000 families are affected by rare diseases in Greece. "The Ministry of Development is implementing targeted actions that strengthen research activity and infrastructure, while promoting cooperation between the private and public sectors," mentioned **Stavros Kalafatis**, Deputy Minister of Development, responsible for Research and Innovation, in his greeting.


**Memi Tsekoura**, President of the Greek Patients' Association, highlighted the need for a binding National Action Plan, the acceleration of registry development, and the assurance of timely and equitable access to innovative treatments, with active patient participation in decision-making. "Rare Diseases, despite their designation, affect a significant part of the population and are accompanied by critical challenges, such as delayed diagnosis, inequalities in access to treatments, and a lack of coordinated care," she stressed, closing her speech.

**Spyros Sapounas**, MD, MSc, PhD, Endocrinologist, Diabetologist, Head of the National Organization for Medicines (EOF), emphasized that EOF has significantly reduced response times, ensuring equitable patient access to medicines. He also underlined the Organization's continuous support for Rare Diseases Greece, noting that despite progress, there is still significant room for improvement. **Eleftheria Tokatlidi**, CEO, Institute of Pharmaceutical Research & Technology (IFET), highlighted IFET's role as a "lifeline" for timely patient access to innovative drugs, noting the significant reduction in procurement times. As she emphasized, the process can be completed in just 5 days, while for more complex imports it takes up to one month, while simultaneously strengthening transparency and achieving lower prices.

In the **Policy Firechat** between **Dimitrios Athanasiou**, Vice President, Rare Diseases Greece, EMA PCWP Member, GPA WDO, Board Member, and **Marios Themistocleous**, Deputy Minister of Health, regarding **Rare Diseases as a National Priority**, Mr. Themistocleous stated, among other things, that "*The National Action Plan has been submitted and is in its final processing stage,"* emphasizing that one of the problems to be solved is equitable access, and especially access to early diagnosis.

**Genomic Medicine and National Strategy**

The example of Australia in genomics and its applicability in Greece was presented by **Professor John Christodoulou**, Director, Genomic Medicine Theme, Murdoch Children's Research Institute, Melbourne, Victoria, Australia, in his introductory speech to the 6th panel, **"The Need for a National PROGRAM: Genetic Care for All."** The session was moderated by **Dimitrios Athanasiou**, MBA, Vice President of Rare Diseases Greece (ESAE), and **Athina Ververi**, Clinical Geneticist-Pediatrician. He stressed the importance of carrier screening and expanded newborn screening, as well as the need for integrating genomic technologies, utilizing artificial intelligence, and stable funding for a more preventive and personalized health system.

In the ensuing discussion, with the participation of **Christina-Maria Kravvari**, Secretary General of Public Health, Ministry of Health, **Pavlina Karasiotou**, Secretary General for Fiscal Policy, Ministry of Economy and Finance, Hellenic Republic, **Spyros Goulas**, Director, Strategic Planning Directorate, EOPYY, **George Papadimas**, Consultant Neurologist, A΄ Neurological Clinic, Athens University, Head, Laboratory of Muscle


Diseases, Aeginetio Hospital, **Christina Kanaka-Gantenbein**, MD, PhD, FMH (CH), Professor of Pediatrics-Pediatric Endocrinology, Director of the 1st Pediatric Clinic and Choremium Research Laboratory of the Medical School NKUA, "Agia Sophia" Children's Hospital, **Periklis Makrythanasis**, MD, PhD, Medical Geneticist, Associate Professor of Medical Genetics at the Medical School of the National and Kapodistrian University of Athens, Researcher, Biomedical Research Foundation Academy of Athens (BRFAA), **Marianna Konstantinidi**, General Manager Greece, Cyprus & Malta, CSL, and **Anthony Aouad**, Sanofi General Manager Greece, Hungary & Ukraine, the need for a structured national strategic plan that integrates genetics into clinical practice was highlighted. It was noted that since 80% of rare diseases have a genetic basis, clear referral protocols, expanded newborn and prenatal screening, access to and reimbursement for molecular tests, and the development of national registries and networks of excellence are required. The speakers concluded that equitable access to genetic care is a critical issue of public health and social justice.

**Strengthening National and European Research Networks**

During the 7th section, **"JARDIN – STRENGTHENING GREECE'S ROLE IN ERNs and CTs (2026-2030),"** in his keynote speech, **Alexis Arzimanoglou**, Coordinator European Reference Network on Rare and Complex Epilepsies ERN EpiCARE - Hospital San Juan De Dios, Barcelona, highlighted European Reference Networks as a mature and efficient system that strengthens diagnosis, treatment, and collaboration in rare disease care. He emphasized that strengthening Greece's participation in ERNs is a significant step for equitable patient access and an opportunity for Greek doctors to actively participate in pan-European collaborative networks.

Subsequently, a panel featuring **Adamantia Englezopoulou**, Alternate National Representative to the Board of EU Member States for Rare Diseases, Manager General Hospital of Corfu, **Labrini Pappa**, Head, Department of Preparedness and IHR, Directorate of Preparedness and Response, National Public Health Organization (EODY), **Georgios Tsivgoulis**, MD, PhD, MSc, FESO, FEAN, FAAN, Professor & Chairman of Second Department of Neurology, School of Medicine, NKUA, "Attikon" University Hospital, Athens, Greece, **Michael Himonas**, General Manager at Hellenic Association of Pharmaceutical Companies, Member of the Board of Directors of EFPIA, and **Evangelia Koraki**, President HACRO (Hellenic Association of CROs), President & CEO CORONIS Research SA, examined how the country's role in European Reference Networks (ERNs) and clinical trials can be substantially strengthened. The discussion focused on increasing Greece's participation in ERNs and the "JARDIN Joint Action," the creation of a national coordination mechanism for ERNs, the role of ERNs as a driver for strengthening Clinical Trials in Rare Diseases, and the strategy for development and international cooperation for the period 2026–2030. In conclusion, the importance of the country's active and coordinated participation in European Networks and initiatives was


highlighted, aiming to improve Care, Research, and access to Treatments for Rare Diseases. The moderators of the session were **Dimitrios Athanasiou**, MBA, Vice President of Rare Diseases Greece, and **Maria Efstratiou**, President of the Pancretan Association of Parents & Friends of Children with Neoplasia "Iliachtida."

The **8th session, "ERDERA & the Greek Research Capacity: Connecting Greece with Europe,"** with speakers **Stelios Kympouropoulos**, Psychiatrist, Former Member of European Parliament, **Christina Kyriakopoulou**, PhD, Policy Officer, Health Research Programmes - Scientific advice: Rare diseases, Health data, AI in health research, **Eleni Grigorakaki**, MSc, MBA, Regulatory Affairs Manager, MSD & Member of the Advisory Board of the European Rare Diseases Research Alliance (ERDERA MAB), **Helen Papadaki**, Professor of Haematology, Dean, University of Crete, School of Medicine, **Dimitris Plexousakis**, Professor in the Department of Computer Science, University of Crete, Head of the Information Systems Laboratory (ISL), Institute of Computer Science, FORTH, and **Antonios Gypakis**, Head, Directorate of Research and Innovation Planning and Programming, highlighted Greece's dynamic presence in the European Research ecosystem and its role in shaping the future of Innovation in Rare Diseases.

The discussion, moderated by **Vasileios Karatzias**, President of Rare Diseases Greece (ESAE) and President of the Hellenic Friedreich's Ataxia Association, and **Kate Theochari**, Vice President of Rare Diseases Greece and President of the Panhellenic Association of Patients & Friends of People Suffering from Lysosomal Diseases "Solidarity," focused on the role of ERDERA as a key European initiative for research into rare diseases, the creation of the National Mirror Group (NMG) and its strategic importance for Greece, the evaluation of the prospect of creating a Greek Research Hub for Rare Diseases, and finally, the connection of national research activity with European Collaborations.

**Data Management and Clinical Assessment Preparation**

The 9th session, **"Data, Orphacodes & Digital Readiness,"** moderated by **Dimitrios Athanasiou**, Vice President, Rare Diseases Greece, EMA PCWP Member, GPA WDO, Board Member, focused on the critical role of Registries, interoperability, and Digital Tools in the management of Rare Diseases. Specifically, the panel discussion between **Georgios Margetidis**, Head of Sector, European Commission Health and Digital Executive Agency, **Panagiota Mitrou**, MD, PhD, Internist - Diabetologist, Lecturer of the University of Athens, Head of Independent Department of Therapeutic Protocols and Patient Registries, Ministry of Health, **Konstantinos Mathioudakis**, Head of Primary Healthcare Systems Department, General Directorate for eHealth (IDIKA S.M.S.A), and **Konstantinos Chalkias**, MD, Internist, Executive Member of the Board of Directors, KETEKNY – Greek DRG Institute, focused on the development of national Rare Disease Registries and their connection with European systems, the need for uniform classification through Orphanet Greece, the full adoption of Orphacodes, and the


enhanced use of tools such as the #CPMS and Registries. They also discussed the use of digital tools like the Rare Diseases Greece Map and the myHealth@myHands Project to improve care.

Participants emphasized that data is a strategic asset for rare diseases, enabling improved diagnosis, monitoring of patient outcomes, and more efficient allocation of healthcare resources. In this context, the alignment of national registries with European data infrastructures was highlighted as a key enabler for research, innovation, and policy effectiveness. The discussion closed by emphasizing the importance of digital readiness and data utilization to support decision-making, research, and equitable patient access to health services.

The final session, **"Access & HTA – Preparing Greece for JCA,"** moderated by **Menia Koukougianni**, Health Project Manager, Association of Rare Diseases of Greece, CO-founder of the NGO KARKINAKI - Information on Cancer in Childhood and Adolescence, focused on the implementation of Joint Clinical Assessment and the conditions for equitable and timely access to treatments. In her keynote speech, **Flora Bacopoulou**, Head, Greek HTA Committee, Professor of Pediatrics-Adolescent Medicine-Clinical Pharmacology, Medical School, National and Kapodistrian University of Athens, highlighted Greece's progress in preparing for the implementation of the Joint Clinical Assessment (JCA) and alignment with the European HTA regulation. She stressed the importance of strengthening the national HTA system, developing methodological guidelines, and the pilot implementation of new procedures. She underlined that the key objective is to ensure equitable and timely access to innovative and cost-effective treatments. Simultaneously, she pointed out the critical role of cooperation between institutions, experts, and patients for the successful implementation of the new framework.

The panel discussion between **Dr. Nandia Gogozotou**, MBA, MSc, PhD, NE President of the National Health Service Agency (EOPYY), President of the Drug Price Negotiation Committee, **Chara Kani**, Pharmacist, Head of the Department of Planning and Monitoring of Drug Administration at the Medicines Directorate, General Directorate of Organization and Planning of Health Services (E.O.P.Y.Y.), **Antonis Moraris**, Oncology Market Access Lead, AstraZeneca Greece, **Haris Plassaras**, Head of Market Access and Government Affairs Greece, Cyprus and Malta, MEDISON Pharma Greece, and **Marieangela Economopoulou**, Head of External Affairs and Patient Value Access - Greece, Cyprus, Malta, TAKEDA, focused on the lessons learned from HTA in Greece and the "WHO Pilot" program, the role of Patient participation, Real-World Evidence (RWE), and system readiness, the country's preparation for the implementation of JCA, and finally, on ensuring equality and timely access to Innovative Therapies. The session concluded with a collective call for the establishment of a modern, transparent, and efficient HTA


framework, aimed at safeguarding and accelerating access to essential treatments for patients with rare diseases.

The conference's Grand sponsor was **Takeda**. Sponsors included **Ardius Pharma** in collaboration with **Orchard Therapeutics** and **PTC Therapeutics,** as well as **AstraZeneca, CSL Behring, GENESIS Pharma, INTEGRIS Pharma, Ipsen, Medison, Roche** and **Sanofi**. Supporters were **Acadia, BioAnalytica, Chiesi, f-anazitisi, Italfarmaco, ORDAID** and **UCB**. Contributors included **Ariti, Brain Therapeutics,** and **Specialty Therapeutics**, while the conference was also supported by **SFEE**.

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