ZEBRA DIAGNOSIS COALITION

EUROPEAN CONFERENCE ON RARE DISEASES & ORPHAN PRODUCTS (ECRD 2026)

Report from the Conference

"Rare Diseases in a Changing & Competitive Europe: Shaping Policies to Address the Unmet Needs of People Living with Rare Diseases"

3-4 June 2026 | O2 Universum Congress Center, Prague | Hybrid Format

Alongside our active participation in the conference, the Zebra Diagnosis Coalition presented its joint research poster, *"Barriers to Early Diagnosis: How Public Healthcare Systems in Europe Shape the Rare Disease Diagnostic Odyssey",* which was well received by attendees and generated meaningful engagement both on-site and among our online community. ECRD indeed offered a timely opportunity to bring our work to a wider audience: drawing on existing literature, namely the EURORDIS Rare Barometer survey, alongside expert insights, our poster maps seven key structural barriers along the diagnostic journey – that contribute to diagnostic delays of nearly five years, 90% of which occur within the healthcare system itself. Yet, beyond presenting these findings, this joint research poster also serves as a platform to highlight the health economic dimension of the diagnostic odyssey and the Coalition's approach to quantifying its costs (or, put differently, the cost of inaction) and to introduce our broader project and next steps to a wider group of stakeholders.

On 3 and 4 June, the Zebra Dx Coalition attended the 13th edition of the European Conference on Rare Diseases & Orphan Products (ECRD), hosted by EURORDIS – Rare Diseases Europe. The conference brought together over 500 in-person participants, alongside more than 300 online attendees, including people living with a rare disease, patient advocates, policymakers, industry representatives, clinicians, regulators, and Member State officials from across the European Union (EU).

Spread across two days, the programme covered a broad spectrum of topics, going from therapies and medical device to development and access to treatments, to timely and accurate diagnosis, research and prevention, as well as evidence-based holistic care, specialised healthcare, technology assessments, and mental health.

Key takeaways for the Zebra Dx Coaltion are:

Beyond networking and knowledge exchange, ECRD 2026 marked the launch of the European Blueprint for Rare Diseases, a multi-stakeholder initiative that sets out to build a unified European roadmap for rare diseases and laying the foundations for a future EU Action Plan. This framework is designed to identify priority policy actions, define targets for monitoring and accountability, and address existing gaps in care, research, and patient support across Europe. The objective to close the diagnosis gap directly resonates with the Zebra Dx Coalition. A gap that, on the ground, translates into unequal access to specialised care,

and stems from seven systemic barriers the Coalition has documented in a poster that we presented at ECRD. *(Access the poster here)*

The opening session set a collective tone: the greatest opportunity in the field of rare diseases today lies in EU-wide collaborative research, knowledge-sharing through the European Reference Networks (ERNs), and ensuring real and equitable access to treatments. National strategies should now show the way forward: guiding patients to the right centre, at the right time. As Kamil Šaško, Minister for Health of Slovakia, underlined: health economic data is essential. With it, we can identify where action has the greatest impact and thus, develop the right policies accordingly. But translating that into real change requires sharing this data widely; because broader public awareness of the scale of diagnostic delays in rare diseases is what also creates (additional) political pressure, and political pressure itself, is what drives financial investment. And at the end of the day, the measure of that investment is crystal clear: how much we improve time to diagnosis.

> *"Share information widely, build public awareness, and create the political pressure needed for decision-makers to act. At the end of the day, it comes down to investment. And how do we measure that investment? By how much we improve time to diagnosis! Because with the right data, we build the right policies."*
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> **Kamil Šaško, Minister for Health, Slovakia**

In the same vein, MEP Vytenis Andriukaitis (S&D, Lithuania) highlighted a distinctly European paradox: scientific excellence is very much present across Europe, yet fragmentation between healthcare structures persists, slowing down the implementation of solutions that already exist for patients. His roadmap: faster diagnosis, broader access to innovation, fully implement the Clinical Trials Regulation, including cross-border coordination across the EU, and better-connect ERNs to significantly shorten the diagnostic pathway for every patient living with a rare disease. MEP Andriukaitis further stressed that digitalisation and multi-country research are indispensable levers for strengthening European competitiveness, and for tackling the inequalities that continue to exist from one States to another within the Union.

Discussions held during Symposium “Navigating the Many Paths of the Diagnostic Odissey”, hosted by UCB directly echoed the mission of the Zebra Diagnosis Coalition. The Aspire4Rare initiative indeed presented a framework built around case studies and, most importantly, seven archetypes. Namely, seven categories of distinct patterns in rare disease diagnostic challenges, designed to move beyond the broad statement that "diagnosis takes too long" and translate complexity into targeted, actionable policy reforms. Crucially, the session underlined the economic dimension of the diagnostic odyssey: delayed diagnosis generates significant costs for healthcare systems, patients, and society alike, through multiple referrals, misdiagnosis, and lost social and professional participation. Hence, earlier diagnosis, as speakers made clear, is not only a medical and ethical priority: it is an economic one.

And this is precisely where our work converges. Indeed, alike Aspire4Rare, the Zebra Diagnosis Coalition recognises that to be actionable - in other words, to genuinely speak to policymakers and Finance

Ministers - advocacy must be grounded in health economic evidence. As such, quantifying the cost of the diagnostic odyssey across different healthcare system archetypes becomes the foundation upon which the right policies can be built. Because without that evidence, the urgency of the diagnosis gap remains invisible to those with the power to act; while patients, in the meantime, continue to bear the consequences with their lives in the balance.

The above discussions confirmed that delayed diagnosis is a predictable system failure, and a costly one. Yet something stood out to us throughout the conference: the true financial burden of the diagnosis odyssey remained under addressed, with health economics failing to receive the prominence it deserves - as illustrated in our poster. And this is precisely the gap the Zebra Dx Coalition is committed to filling. Mapping these costs across different European healthcare systems, thereby enabling us to identify where policy action can have the greatest impact, remains the basis of our work and, ultimately, what we want to leverage to make the case to those who control the budgets.