Zebra Diagnosis Coalition
Health Economics for RD Dx

ZEBRA DIAGNOSIS COALITION

WORLD ORPHAN DRUG CONGRESS EUROPE 2026

Report from the Conference

26–28 October 2026 | Amsterdam | In-person

The Zebra Diagnosis Coalition recently took part in the World Orphan Drug Congress Europe 2026 (WODC).

In line with our core mission to accelerate earlier diagnosis and timely access to care for people living with rare diseases, by combining health-economic evidence that quantifies the cost of inaction with coordinated policy advocacy at national and European levels, we participated in two sessions:

- A workshop organised by Sanofi *"Early diagnosis: what's needed to make change happen?",* featuring notably Gulcin Gumus (EURORDIS-Rare Disease Europe), Maurizio Scarpa (MetabERN) and Vinciane Pirard (Sanofi), attended by 70+ participants.
- Our own panel "*Zebra Dx Coalition: "How early diagnosis can help with Health Economic outcomes",* attended by 40+ participants, and composed of Gulcin Gumus (EURORDIS-Rare Diseases Europe), Malwina Mejer (Copenhagen Economics), Vinciane Pirard (Sanofi), and Anna Lawson (Alexion - AstraZeneca Rare Disease). Discussions notably focused on: (1) why early diagnosis of rare diseases matters, (2) what are the barriers that stand in the way, and (3) how our national studies work can and will generate the health-economic evidence needed to drive policy change (to accelerate timely diagnosis), and why it is essential to act now.

Key takeaways from the Zebra Dx Coalition - Sanofi's Workshop, WODC 2026 **"*Early diagnosis: what's needed to make change happen*?" |** October 26, from 14:00-17:00

Engaging early with the upcoming Italian Presidency of the Council of the EU

With its new *National Rare Diseases Plan 2026-2029*, the Italian government has made rare diseases a core public health priority to reduce territorial inequalities at the national level. As the country will be taking up the Presidency of the Council of the EU in 2028 H1, we believe this national commitment can translate into concrete momentum across Europe. We are therefore determined to engage ahead with Italian stakeholders to help ensure early diagnosis features on the Presidency's agenda.

Strong convergence with other initiatives across the rare disease community

Initiatives such as HLM4RARE, which examines the barriers along the diagnostic journey, are tackling the same challenges as the Zebra Dx Coalition from complementary angles. In this context, we intend to strengthen our collaboration with such initiatives and reinforce the alignment of our messaging to amplify the common call for earlier diagnosis. We are particularly honor to participate in HLM4RARE's upcoming Conference taking place from 1-3 December 2026, where we will present our initiative and, most importantly our national studies.

The unfortunate absence of HTA bodies from the debate on diagnosis

Health Technology Assessment (HTA) bodies play a crucial role in national healthcare funding decisions across the EU. Yet, they are currently absent from the debate on how to accelerate timely diagnosis of patients living with rare diseases. As a result, arguments in favor of earlier diagnosis at national levels are unlikely to translate into concrete investments. This is precisely where the Zebra Dx Coalition can make a difference: by quantifying the cost of delayed diagnosis at the national level.

Feeding national evidence into JARDIN

The European Reference Networks (ERNs) pool rare disease expertise across the EU, yet their overall impact on diagnosis depends on how well they are integrated into national health systems; and this is where the JARDIN Joint Action has a key role to play. By mapping where, how, and at what cost delays occur along diagnostic pathways, starting with three countries, our national studies aim to provide evidence that can help improve coordination between healthcare systems within the EU.



Sanofi's Workshop, "Early diagnosis: what's deeded to make change happen?" | Monday 26 October, WODC 2026.



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Key takeaways from the Zebra Dx Coalition's Panel, WODC 2026 ***"How early diagnosis can help with Health Economic outcomes"*** | October 27, from 14:10-15:10

Bringing together patient, industry and health economics perspectives, the discussion started from *what* diagnosis means for people living with a rare disease, before turning to the barriers that delay it, the value of our national studies, and *how* this economic evidence can be translated into advocacy at both national and European levels. Three key messages emerged from the panel, each building on the previous one:

- Build health economic evidence to make earlier diagnosis a policy priority
- Quantify the costs of selected barriers to diagnosis across national healthcare systems
- Translate national evidence into coordinated advocacy at national and EU levels

> *"Policymakers need to recognise that leaving rare diseases undiagnosed has a cost, and addressing barriers to diagnosis is an investment for patients, health systems and society."*
>
> **Malwina Mejer** (Senior Economist, Copenhagen Economic), **WODC 2026**

Build health economic evidence to make earlier diagnosis a policy priority

In each country across Europe, the diagnostic odyssey remains long for patients living with a rare disease, while policymakers continue to underestimate the problem. By generating national-level evidence on the economic consequences of having diagnostic delays, particularly looking at costs beyond the healthcare system and at the patient's perspective together, the Zebra Dx Coalition aims to demonstrate *why* earlier diagnosis warrants investment against competing priorities. Investing in rare diseases is ultimately an investment in health systems, with spillover effects for patients, their families and society at large.

Quantify the costs of selected barriers to diagnosis across national healthcare systems

The way a healthcare system is organised largely explains the diagnostic odyssey. Building on the seven barriers identified in our White Paper, our national studies will examine *how* selected barriers operate in three countries (with different healthcare systems), the delays they create and *what* they cost. By covering the whole care pathway, from first symptoms to diagnosis to follow-up, this evidence should help policymakers to see which barriers generate the highest avoidable costs and therefore where action would deliver the greatest value.

> *"We will develop health economic evidence by disease, by country and by barrier, to inform decisions that can be taken within a health system."*
>
> **Anna Lawson** (Alexion - AstraZeneca Rare Disease), **WODC 2026**

Translate national evidence into coordinated advocacy at national and EU levels

Since health remains a national competence, progress on earlier diagnosis must start at country level, and what works in one country may not work in another, particularly in decentralised systems such as Italy. This is precisely where the Zebra Dx Coalition can make the difference: our national studies work, should support country-specific advocacy while feeding into wider EU discussions through extrapolation of national results. Patient organisations are therefore essential partners from the outset, ensuring the evidence reflects patient experience and quality of life on ground, and translates into meaningful policy change, at both national and European levels.



The Zebra Diagnosis Coalition's Panel, " How early diagnosis can help with Health Economic outcomes?" | Tuesday October 27, WODC 2026.

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