Zebra Diagnosis Coalition

BARRIERS TO EARLY DIAGNOSIS

HOW PUBLIC HEALTHCARE SYSTEMS IN EUROPE SHAPE THE RARE DISEASE DIAGNOSTIC ODYSSEY

Despite advances in research and care, many Europeans with rare diseases face diagnosis delays or remain undiagnosed

People living with a rare disease in Europe still face **diagnostic delays of nearly 5 years**, despite advances in genomics, improved understanding of disease mechanisms, a growing number of treatment options, and European collaboration. **Late diagnosis worsens outcomes and generates substantial costs for patients, healthcare systems and society.** While rare diseases are clinically heterogeneous, diagnostic delay is not random. It reflects the organisation of public healthcare systems. This poster maps the typical diagnostic journey and highlights where structural barriers **(B)** arise at each step.

WHY THIS MATTERS

Delayed diagnosis worsens patient outcomes and generates avoidable costs for healthcare systems and society

7x higher medical costs before diagnosis for suspected rare disease patients

(Glaubitz et al., 2025)

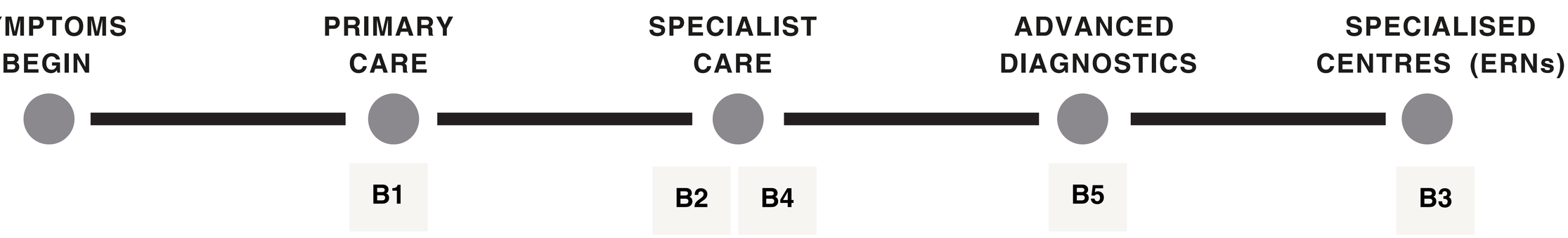
6x higher annual healthcare spending across a patient's lifetime for people living with a rare disease

(Wilsdon et al., 2024)

90% OF DELAYS OCCUR WITHIN THE HEALTHCARE SYSTEM

Methodology: We rely on relevant literature, the EURORDIS Rare Barometer survey, and in-depth interviews with industry experts to better understand where and how barriers arise along the diagnostic journey.

DIAGNOSTIC JOURNEY (SYMPTOMATIC PATIENT)



B1 LIMITED CAPACITY IN PRIMARY CARE

Most **GPs encounter only a single patient with a given rare disease in routine practice**. Limited exposure and training can lead to missed signals and delayed referrals.

B2 FRAGMENTED ORGANISATION OF SPECIALISED CARE

Specialisation into single disciplines disadvantages patients with multisystem rare diseases, contributing to **an additional 3.5 years of delay within the healthcare system**.

B3 POOR COORDINATION WITH SPECIALISED RARE DISEASE CENTRES

Despite specialised expertise, patients typically reach centres of excellence only after **years of inconclusive care**, limiting their impact on diagnostic timelines.

B4 PRIORITISATION OF URGENCY OVER CLINICAL COMPLEXITY

Urgency-based prioritisation delays diagnosis for non-acute rare diseases, with transient or invisible symptoms **adding 1.5 years to healthcare system delays**.

B5 INCONSISTENT AND LATE USE OF GENETIC TESTING

Although 72 per cent of rare diseases have a genetic origin, genetic testing is often deployed late and inconsistently, **adding up to 2 years to the diagnostic journey**.

B6 LIMITED ACCESS TO TREATMENT

Diagnosis can be up to 4 times faster when a targeted EMA-approved treatment is available at symptom onset.

(Wilsdon et al., 2024)

B7 FRAGMENTED INFORMATION SYSTEMS

Only **3 EU countries routinely share patient data across HCPs, and 8 fully use EHRs across care levels**. A limited number have implemented structured rare disease coding (ICD-11 or ORPHAcodes).

Delayed diagnosis of rare diseases is a **predictable system failure that drives avoidable healthcare costs**. Barriers are similar across Europe, but their extent and costs differ between countries. The next step is to document these costs and identify where policy action can have the greatest impact.

Zebra Diagnosis Coalition Members: Alexion, AstraZeneca Rare Disease, argenx, Chiesi, Kyowa Kirin, and Sanofi.

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