Recommendation 2:

Earlier, faster and more accurate diagnosis of rare diseases through better and more consistent use of harmonised standards and programmes across Europe, new technologies and innovative approaches driven by patient-needs.

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This are our main goals

- **All people living with a rare disease known in the medical literature will be diagnosed within six months of coming to medical attention.**

- **All people will have access to the most effective diagnostic technologies, best practices and programmes (including screening) without discrimination and regardless of where they live in Europe.**

- **All currently undiagnosable individuals will enter a European and globally coordinated diagnostic and research pipeline.**

EARLIER, FASTER, MORE ACCURATE DIAGNOSIS

How it works:

1. **Promote equality in access to diagnostic opportunities for people living with a rare disease or suspected rare disease, no matter where they live.**

2. **Enable patients to navigate health systems with ease, following the most appropriate and direct route to obtaining a diagnosis, to connect with others in similar situations, and learn how to best manage their disease and participate in research in a safe and timely manner.**

3. **Co-design - with healthcare professionals and patients - care pathways to most efficiently guide people living with a rare disease from diagnosis to highest quality care and, where possible and appropriate, to the most relevant European Reference Network.**

EARLIER, FASTER, MORE ACCURATE DIAGNOSIS

How it works:

4 **Improve diagnostic expertise by fostering European and global networking of highly specialised healthcare providers and by ensuring greater interoperability and standardisation of data able to support diagnostics whilst ensuring particular transnational collaboration to diagnose the most complex presentations, rarest diseases and undiagnosed cases.**

5 **Ensure an integrated, international approach to patients with currently undiagnosable conditions, ensuring the absence of diagnosis does not preclude access to the best possible care and support.**

6 **Raise awareness on rare diseases in medical curricula and amongst all primary/ front-line health and social care professionals and specialists. Foster broad and equitable implementation of next generation sequencing and other emerging new technologies into national healthcare systems, to facilitate and speed up access to diagnosis.**

HOW TO ACHIEVE THIS?

- **GUIDING**: A clear, systematic and European-wide (indeed sometimes global) approach to rare disease diagnostics must be ensured, founded upon the ability to guide patients towards centres of expertise or equivalent.
- **SHARING:** access transnational diagnostics platforms, and capture - and systematically manage - data on patients for whom a diagnosis is not forthcoming.
- **SUPPORT:** Continued support must be ensured for multinational and multistakeholder research linking omics data, clinical data and biomaterials with well-defined patient cohorts and applying them in the clinic, building on the work of existing initiatives such as the European Joint Programme on Rare Diseases and Solve-RD.
- **GUIDELINES:** Existing and future best practice guidelines to support the diagnosis of rare diseases (such as decision trees and patient pathways) should be visible and findable at the European level (via European Reference Networks ERNs and Orphanet).
- **INVENTORYING:** The Orphanet services pertaining to diagnostics (resources concerning the definition and inventorying of diseases, and the database on expert clinical centres and laboratories) should increasingly be co-created and co-curated together with ERNs, and should be sustained by European action.
- **FUNDING:** Funding bodies in Europe and all other world regions should target diagnostics for subpopulations, indigenous people, and other culturally and linguistically diverse populations in a culturally safe manner (including populations in developing nations): this will support the genetic and phenotypic characterisation of rare disease populations to enlarge patient cohorts and advance knowledge and understanding.
- **RESEARCH:** Research should be fostered at European level to elucidate the determinants of the heterogeneity across EU Member States in terms of diagnostic performance. To conduct a cross-border health economics assessment of diagnostic and screening technologies, comparing costs and benefits relative to those currently incurred under the diagnostic ‘odyssey’.

LINKING BETTER DIAGNOSTICS TO CARE PATHWAYS

- **Countries should define clear national strategies to support RD diagnostics and should support professionals involved in diagnostics -and through them, patients- in their national territory to access specialised diagnostic platforms; in particular, to utilise genome-phenome platforms and similar tools.**
- **Countries should adopt, and provide the means (financially and organisationally) to actually implement EU level best practice recommendations on diagnosis and screening.**

FROM THE BEGINNING: NEWBORN SCREENING

- **The proposed activities highlighted in the EUCERD Opinion on Newborn Screening should be revisited and implemented through a European-level body or programme; new solutions proposed in EURORDIS Key Principles for Newborn Screening should be considered.**
- **Countries should work collaboratively to share best practices and HTA data concerning newborn screening programmes.**
- **The cost-effectiveness of newborn screening should be calculated and set against the costs of the diagnostic odyssey and costs to the health and social systems in the absence of an accurate diagnosis.**

LINKING BETTER DIAGNOSTICS TO CARE PATHWAYS

AND AFTER IT'S THE ROLE OF THE PRIMARY CARE

- **Individuals with suspected diagnoses must be referred to the most relevant specialist centres/ centres of expertise / coordination hub at the earliest opportunity: the precise role which ERNs could play in facilitating a diagnosis for rare disease patients lacking one should be clarified and better implemented at the national level.**
- **As triage to identify a possible area of specialism for referral rests upon awareness in primary and secondary care of how and where to access such expertise, national referral pathways to tertiary centres of expertise (or a catch-all coordination hub/centre most efficiently accompany people living with a rare disease from diagnosis to highest quality care and where possible to the European Reference Network (ERN) covering the disease. for rare diseases) must be elucidated and made publicly available.**

BUT ALWAYS IT WILL BE UNFORTUNATELY PATIENTS WITHOUT DIAGNOSIS

- **Countries should build knowledge on existing undiagnosed rare disease patient populations – research should be conducted to establish number of undiagnosed patients, and socio-economic impact including impact on patients' and families' quality of life and ability to access health and social care.**

WHAT DO PEOPLE LIVING WITH A RARE DISEASE THINK?

The following figures come from several Rare Barometer surveys carried out in Europe between 2016 and 2021. In these surveys, people living with a rare disease told us their views on a range of issues, such as participation in research, access to treatments, social care needs, the impact of COVID-19 on their lives and their hopes for 2030. Find out more: eurordis.org/voices

GET A FASTER DIAGNOSIS IF DISORDERS ARE KNOWN IN THE MEDICAL LITERATURE

97% of people living with a rare disease agree to share their health data in order to foster research. This dataflow (well analysed, securely stored and constantly updated) is central to build a robust and up-to-date medical literature and to coordinate diagnosis techniques across the globe.

ACCESS TO MORE EFFECTIVE DIAGNOSIS STRATEGIES: NEW-BORN SCREENINGS

94% of respondents support the diagnosis of rare conditions at child's birth, pushing for implementing new-born screening across Europe.

People living with a rare disease also favour the use of new technologies to diagnose rare conditions before birth: either during pregnancy (87%) and around the time of conception (80%).

GET THE BEST DIAGNOSIS, REGARDLESS OF WHERE ONES LIVE, THANKS TO THE EUROPEAN REFERENCE NETWORK

+ The centers of expertise in rare diseases put in place in Europe (European Reference Network) are already showing results in significantly reducing the time between the first sought medical advice and the confirmed diagnosis of a rare or complex disease: it currently takes 2.8 years in ERNs to diagnose while it takes 4.1 years elsewhere in Europe.
+ ERNs also perform more genetic testing than non-ERN HCPS to either get or confirm a rare/complex disease diagnosis.