

Mapping of Resources from Networks for Undiagnosed and Newly Diagnosed Ultra-Rare Diseases

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ABSTRACT

Every year, 65000 children across Europe are born with an undiagnosed disease (1). The diagnostic odyssey lasts 5 years on average for a rare disease patient (2). This delay in diagnosis prevents patients from accessing proper care and treatment in a timely manner (3). Several national and international initiatives have been established to improve the lives of people with undiagnosed rare diseases and their families. However, currently, there is not a comprehensive mapping of resources for undiagnosed or newly diagnosed Ultra-Rare diseases.

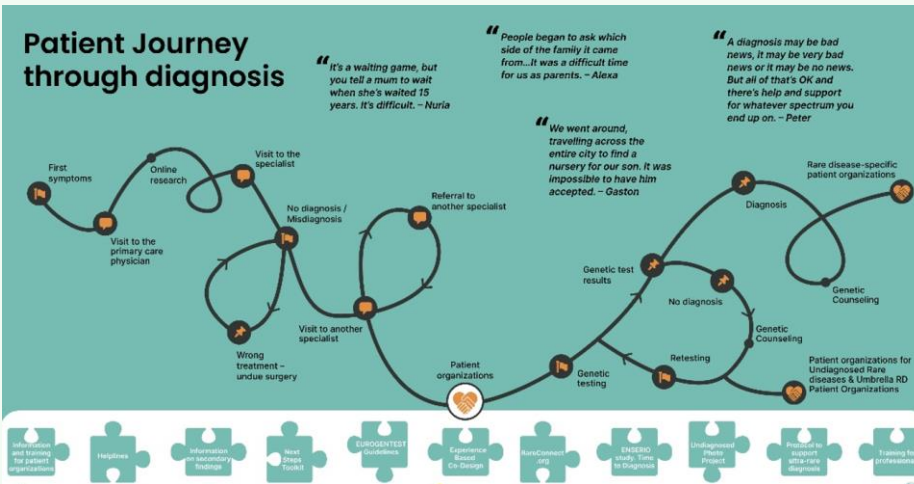
To date, Solve-RD project, where 8463 datasets (8152 WES and 311 WGS) including 5205 affected individuals from 4862 families are being analyzed, have solved 130 rare disease cases by the re-analysis of unsolved exomes. Within this project, EURORDIS initiated the Community Engagement Task Force (CETF), a multi-stakeholder community of patients, scientists, and clinicians, to support the needs of the undiagnosed and recently diagnosed patients and leave a legacy of a strengthened undiagnosed

METHODOLOGY

Key stakeholders active in the field of diagnosis within the national and international initiatives were invited to become members of the CETF. These include the Undiagnosed Diseases Network International (UDNI), SWAN (Syndrome Without A Name) Europe, European Patient Advocacy Groups within the European Reference Networks and The Global Commission to End the Diagnostic Odyssey for Children.

CETF members gathered through three teleconferences and a face to face meeting over 15 months where they discussed issues addressing specific needs of undiagnosed rare disease patients and their families such as lack of information for medical professionals and the need for standardization when receiving a diagnosis. Resources from members (including toolkits, surveys, fact sheets and illustrations) were compiled to develop a roadmap.

Figure2. Diagram representing the patient journey through diagnosis and resources from CETF member organizations



RESULTS

Figure1. Members of the CETF

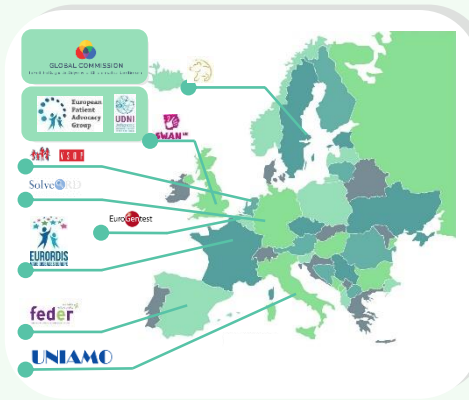


Figure 3. Highlights on some key resources from CETF member organizations

Protocol to support ultra-rare diagnosis

Developed by AnDDI-Rares, this document aims to support genetic services caring for families faced with an ultra-rare diagnosis. To better address families' expectations, it is important to prepare them before NGS testing, at the time of returning the results and also later on.

ENSERIO- Analysis of the reality of the group of people without diagnosis

This study is conducted by FEDER and focuses on the psycho-social and professional impact of living with an undiagnosed disease by analyzing quantitative data from 78 people who are living with an undiagnosed condition.

Undiagnosed Photo Project

This Wilhelm Foundation initiative aims to reach a diagnosis through sharing and it is created for patients who have been evaluated at the Undiagnosed Disease Program (UDP) and are still undiagnosed after the program.

CONCLUSION

Regular and meaningful engagement of representatives within and across key initiatives represent the start of an active undiagnosed community. Importantly, patients as equal and valued partners are driving forward active ongoing collaborations among the different stakeholders.

ACKNOWLEDGEMENTS & REFERENCES

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- Introducing SWAN Europe (press release) April 2017 Genetic Alliance UK <https://www.undiagnosed.org.uk/news-event/introducing-swan-europe/>
- International Joint Recommendations to Address Specific Needs of Undiagnosed Rare Disease Patients <https://www.eurordis.org/publication/international-joint-recommendations-address-specific-needs-undiagnosed-rare-disease-patients>
- European Organisation for Rare Diseases. Survey of the delay in diagnosis for 8 rare diseases in Europe (EurordisCare2). EURORDIS Fact sheets, 2007. <https://www.eurordis.org/publication/survey-delay-diagnosis-8-rare-diseases-europe-2-eurordiscare2/>

