

State of the Art for Rare Disease – Activities in EU Member States and Other European Countries

Malta Report

Definition of a Rare Disease

Malta has adopted the European Commission definition of a rare disease. More information on this can be found at <https://health.gov.mt/en/dhir/Pages/Registries/Rare-diseases.aspx>.

National Plan

Malta has not adopted a National Plan or Strategy for rare disease. An internal action plan serves as a basis for a number of specific actions taken by the Ministry. These actions include:

- The establishment of the Rare Disease Register
- The creation of the Treatment Abroad Service – the provision of tertiary specialised care abroad to a substantial number of rare disease patients.

Organisation of Rare Disease Health and Social Care

Centres of Expertise

There is no official policy for the designation of Centres of Expertise for rare disease in Malta. No formal mechanism has yet been identified and the final mechanism will depend upon the final ERN structure. Malta is working towards the establishment of a formal process of designation for both Centres of Expertise and affiliated centres so that they can participate in ERNs as defined by the EU.

European Reference Networks (ERNs)

As above, Malta is working towards establishing a formal process of designation for both Centres of Expertise and affiliated centres to participate in ERNs. To date no Health Care Provider in Malta has been able to form part of an ERN application, either as a coordinator, member or affiliated member.

Rare Disease Registration

In Malta national rare disease registries are in place. No specific legislation was enacted in this regard until September 2016. All epidemiological registers are kept within the remit provided by data privacy legislation for the protection of public health and management of health services.

As of September 2016 the status quo of rare disease registration in Malta was as follows.

The number officially registered:

- 2,235 persons suffering from rare diseases have been coded (assuming a rare disease population of 27, 500 in Malta this represents 8%). The process is dynamic and open-ended as people die, diagnosis criteria change, definitions of rare diseases change. Malta continues to update this information regularly.
- 442 different diagnoses have been identified within the population, estimate were that >600 types would be found in Malta.
- Treatment abroad with the highest diagnostic yield are to be expected.

Data is collected from the following sources:

- Congenital Anomalies Register (completed)
- Cancer Registry (completed)
- Treatment Abroad List of Patients (completed)
- Rare diseases referred from website (ongoing; needs revamp)
- Clinicians' registers (to be started – no dates yet; possibly still data protection, legal and other administrative issues pending)

- Genetics data (to be started – no dates yet; possibly still data protection, legal and other administrative issues pending)
- Pathology Database to be started – no dates yet; possibly still data protection, legal and other administrative issues pending)

Malta's immediate plans are:

- To further populate the register
- Urgent need for IT support – the creation of software between existing systems
- Iron out the remaining data protection, legal administrative issues (esp. Ownership of data, data sharing at what level etc.)
- Collaboration with NGOs

There is no specific funding for the rare disease registries as they all form part of the Health Information Departmental Services. Approximately there are 2235 patients currently registered.

With regard to data, Orphacoding is used within the registries and Malta is currently in the process of trying to integrate data from within other internal sources like hospitals, pathology laboratories and genetic laboratory data.

Neonatal Screening

No legislation or written policy currently exists pertaining to neonatal screening in Malta. The Neonatal programmes began following a directive from the Ministry of Health. The programme screens for two diseases: Neonatal Congenital Hypothyroidism and Haemoglobinopathies.

Guidelines and Training Activities

Clinical Practice Guidelines

Practice Guidelines are typically produced, in Malta, to define drug entitlement. Malta has in place national policies for the development, adoption and implantation of Clinical Practice Guidelines.

Training and Education

A symposium is held every year, at the end of February, between the DHIR and the University of Malta.

Information Resources for Rare Diseases

Orphanet Activities

Malta does not currently have a national Orphanet team. The national representative on the CEGRD is the same person who is working to undertake all functions within an Operational National Orphanet team.

National Helplines

Currently there is no helpline in place dedicated to rare diseases in Malta. There are plans to set up a helpline, this will form part of the collaboration between the NGOs and the Ministry. A helpline does exist for those patients who will be accessing treatment services abroad.

Official Information Centres

There are no official or unofficial information centres for rare disease in Malta.

Rare Disease Research Activities

Existence of rare disease programmes/projects

Malta does not currently have in place any specific programme or projects to fund or facilitate rare disease research. There are also no plans for such research in the future nevertheless a cost benefit assessment has been carried out.

Participation in E-Rare and International Research Initiatives

At present Malta does not participate in either E-rare or IRDiRC due to resource restrictions.

National Alliance of Patient Organisations and Patient Representation

There is a national alliance for rare disease patient organisations in Malta. Since February 2014 the cause of rare diseases in Malta has featured prominently on the national agenda, particularly due to the work of the Marigold Foundation. The foundation began a two year long campaign which sought to highlight the need for a national alliance for rare diseases in Malta. Through its fund-raising efforts The Marigold Foundation raised enough funds to set up the National Alliance for Rare Diseases Support – Malta. Co-founder of the Alliance, Chairperson of the Marigold Foundation and spouse of the PM, Michelle Muscat started working closely with EURORDIS, establishing a link that has served to promote rare diseases both on the national and international agenda. Mrs Muscat has been honoured as European patron of EURORDIS for the last three years and was chosen to co-chair the EURORDIS Black Pearl Event alongside Princess Ann de Ligne of Belgium.

Through the establishment of the National Alliance for Rare Diseases, which brings together all the stakeholders including patients and their relatives, researchers, medical professionals and other organisations representing specific rare diseases, representation of patients has been magnified with more care, support and awareness for the benefit of the patients and their relatives.

The National Alliance and its founder, The Marigold Foundation, worked incessantly so that a National Directory could be started in collaboration with the Ministry of Health. This National Directory was launched in 2016. This National Directory will make it possible to trace rare conditions and diseases on the small island state of Malta, where rare is even rarer.

Integration of Rare Diseases to Social Policies and Services

Specific actions exist to enable real access for people with rare diseases to general social/disability programmes. Actions are limited to those provided by the Ministry of Social Affairs, multidisciplinary groups exist within the commoner specialities rather than just limited to a specific diseases.

There are specific measures in place to support the integration of rare disease specificities into the national system responsible for assessing a person's level of functioning. In Malta, for example, there is a specific employment programme for persons with a disability but not specific for rare diseases.

Rare Disease Day

The Marigold Foundation together with the National Alliance for Rare Disease Support – Malta, has celebrated Rare Disease Day and Rare Disease Month 2016, with various events including lectures at the University of Malta, patient gatherings, fund-raising dinners, art and cultural expositions and seminars and lectures.

Since its establishment, the Alliance together with the Ministry of Health and the University of Malta, organised various events to raise awareness both at the national and international level and is currently working to raise the rare disease issue at the European Presidency during Malta's tenure. The Alliance is also working closely with EURORDIS and RDI to raise the agenda of rare diseases on a United Nations platform.

Other

Since 2014 Malta has accomplished several rare disease related achievements:

- The introduction of a new updated neonatal congenital hypothyroid screening programme
- The establishment of the rare disease registry.

Information on Genetic Testing and Orphan Medicinal Products will be available in the next update.