

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

Sweden Report

Definition of a Rare Disease

Sweden has not adopted the European Commission definition of a rare disease. Instead Sweden chooses to define rare disease as having a prevalence of no more than 1 patient per 10,000 persons combined with a severe lifelong disability.

National Plan or Strategy

Sweden has not adopted a National Plan or Strategy for rare disease.

In June 2010, the Swedish Government decided to establish a national focal point for coordination in the field of rare diseases, a €350,000/year project, with the main objective of coordinating rare disease efforts and disseminating knowledge and information within and between health services, NGOs and other stakeholders. The decision represented an important step towards a better use of the resources available for patients with rare diseases and the patients' relatives. The National Board of Health and Welfare was commissioned to establish this national focal point. At the end of 2011 the National Board of Health and Welfare announced the new National Function for Rare Diseases (NFSD - Nationella Funktionen för Sällsynta Diagnoser). Their work includes the:

- Promotion of coherence and the coordination of health care resources for people with rare diseases
- To accomplish an increased cooperation with the social insurance, employment services, social services, NGOs and other actors.
- They also contribute to the dissemination of knowledge and information and to the exchange of good practice and experiences. NFSD started on 1 January 2012 and the assignment has been entrusted to the non-profit rare disease care facility Ågrenska. An inventory of available resources for people with rare diseases was one of the first tasks for the NFSD.

The Swedish Government decided in October 2011 to assign the National Board of Health and Welfare the task of developing a national strategy for rare diseases. The National Board of Health and Welfare have worked together with the NFSD and other stakeholders to develop the strategy. In October 2012, the Swedish national strategy for rare diseases was transmitted to the government. In 2016, the plan had still not been officially adopted.

Organisation of Rare Disease Health and Social Care

Centres of Expertise

There is an official policy in place in Sweden for the designation of Centres of Expertise for rare disease. This national policy has been fully implemented and the objectives and criteria for the centres have been developed at the national level based on the EUCERD criteria.

Currently there are 62 Centres of Expertise complying with the National Policy, the number of Centres in Sweden divided by million inhabitants is 6 and the number of Centres fulfilling the EUCERD criteria is 15. The Centres of Expertise criteria ensure a holistic approach and work is currently underway to develop the Centre activities.

European Reference Networks (ERNs)

Sweden has a formal process in place for the endorsement of Healthcare Providers to participate as members or coordinators within a European Reference Network.

Currently there are 6 HCPs participating as full members of 19 ERNs in Sweden.

Rare Disease Registration

Neither national nor regional RD registries exist, but disease specific registries are available in Sweden. Rare disease registries are under the same laws and regulations as any other registry. The current rare disease registries are as follows:

- Svenska Hemofiliregistret (Hemophilia A&B and von Willebrand disease) <http://www.kvalitetsregister.se/hittaregister/registerarkiv/blodarsjuka.2406.html>
- Medfödda metabola sjukdomar (metabolic disease) <http://rmms.se/>
- Svenska CF register (cystic fibrosis) <http://www.kvalitetsregister.se/hittaregister/registerarkiv/cystiskfibros.2317.html>
- Svenska barnpilepsiregistret (epilepsy, children) <http://kvalitetsregister.se/hittaregister/registerarkiv/barnpilepsi.2370.html>
- The Swedish registry of congenital heart diseases, SWEDCON <http://www.ucr.uu.se/swedcon/>
- PID-care (primary immunodeficiency) <http://www.pidcare.se/>
- Svenska Neuroregistret (Myastenis gravis, Narkolepsy, Inflam. polyneuropathy, ALS) <http://www.neuroreg.se/>
- Neuromuskulära sjukdomar i Sverige (Dystrofinopathy, LGMD,FSHD,CMD,Dystrofia myotonica, Kongenital myopathy, Welander myopathy,metabolic myopathy, SMA) <https://nmisse.wordpress.com/>

Genetic Testing

Genetic testing is performed at the Department of Clinical Genetics of each University hospital. WES and WGS are in some cases carried out in close collaboration with Science for Life Laboratories.

Genetic testing abroad is possible and is widely used, and there are not any specific regulations opposing this.

Neonatal Screening

All parents are offered screening of their new-borns for the diseases listed below within 48 hours. The blood sample is stored in a biobank if the parents give their consent.

Currently the 24 tests included in the neonatal screening programme are as follows:

- Phenylketonuria,
- Hypothyroidism,
- Congenital adrenal hyperplasia,
- Biotinidase deficiency and galactosemia,
- MCAD deficiency,
- LCHAD deficiency and other defects in TFP,
- VLCAD deficiency,
- Dysfunction of the carnitine cycle molecules CPTI, CPTII and CACT,
- Primary carnitine deficiency CUD,
- Isovaleric aciduria,
- Methylmalonic aciduria MMA,
- Glutaraciduria type I and 2,
- Betaketothiolase deficiency,
- Citrullinemia,
- Argininosuccinate lyase deficiency (ASA),
- Arginase deficiency,
- Maple syrup urine disease (MSUD),
- Tyrosinemia type 1,
- Propionic academia
- Homocystinuria.

Guidelines and Training Activities

Clinical Practice Guidelines (CPGs)

Sweden's health care system is decentralised and run by 21 county councils/regions, mainly responsible for CPGs. Clinical Practice Guidelines in the rare disease area are available on [nfsd.se](http://www.nfsd.se):

<http://www.nfsd.se/om-sallsynta-diagnoser/Vardprogram-och-riktlinjer/Sammanställning-av-wardprogram-och-riktlinjer/>

Training and Education

Since 2011 residency doctors have participated in a five days program at Ågrenska, for families having children with a rare disease. The main objectives of the course is to increase knowledge concerning rare disease, insight of the impact of the disease on the whole family, knowledge about the siblings' relation, knowledge about talking to the parents and children about serious topics, knowledge about official support from the society and possibilities to reflect before and after the course.

During the Ågrenska Family Programs, the RD family professional network had the opportunity to participate in two lecture days to get more knowledge about the disease and the needs of the patient.

- Educational events are regularly organised at the Karolinska Centre for Rare Diseases.
- Specialist courses for medical doctors within RD have been organised at the Karolinska Centre for Rare Diseases.
- Skeletal dysplasia workshops have been organised by several different clinics at the Karolinska University Hospital.
- Dysmorphology and syndrome diagnostic workshops for medical doctors have been organised at the Department of Clinical Genetics Centre for Rare Diseases at the Karolinska University Hospital.

Information Resources for Rare Diseases

Orphanet Activities

Sweden has an operation National Orphanet team which is housed within the Karolinska University Hospital. Orphanet - Sweden (orphanet.se) has a national website, which is directly linked to the international Orphanet portal.

Different RD documents and policies can be found on this site as well as relevant information regarding RD expert centres and current educational and information events in Sweden. Description of the national team, contact and work procedures is also available.

Orphanet - Sweden has several national collaborations.

One of the major achievements during the last two years is the direct links that have been created between the national RD databases (IOD) and the international Orphanet portal. On one hand Orphanet is presented on the front page of the national database as a complementing source of RD information. On the other hand all Swedish and English texts that are produced by RD experts in Sweden are directly linked to the corresponding page on the Orphanet portal. In total, 1000 have been created.

Orphanet - Sweden is also a member of the National Communication and Information Group, which was established by NFSD to harmonise RD information. One of the completed projects focused on collecting and updating information regarding RD Patient Organisations in Sweden, which was carried out in collaboration between NFSD, IOD, Orphanet and RD alliance.

Orphanet-Sweden has been very active to promote formation of expert teams within Centre of Expertise according to the EUCERD criteria. A large number of teams have been then able to participate as core members in ERNs within their area of expertise.

Orphanet-Sweden is involved in the efforts to include Orphacodes in the currently developed RD registries.

National Helplines

Sweden does not currently have a helpline for rare diseases however discussions are ongoing.

Official Information Centres

There are official and unofficial information centres for rare disease in Sweden. These centres are organised and financed as follows:

- The Swedish National Agency for rare diseases, financed by the National Board of Health and Welfare. <http://www.nfsd.se/>
- The Swedish Information Centre for Rare Diseases, is seated at the University of Gothenburg. The Swedish National Board of Health and Welfare finance the Centre. <http://www.socialstyrelsen.se/ovanligadiagnoser>
- Orphanet Sweden, organized by Karolinska University Hospital, financed by RD-action and Karolinska University Hospital, Centre for Rare Diseases. <http://www.orphanet.se/national/SE-SV/index/hemsida/>
- Swedish National Orofacial Resource Centre for Rare Diseases, Mun-H Center, financed by the Swedish National Board for Health and Welfare and the Public Dental Service, Region Västra Götaland, Sweden. <http://www.mun-h-center.se/sv/Mun-HCenter/>
- Ågrenska Foundation, a non-profit national competence centre. Partly funded by grants from the state and fees from the counties. www.agrenska.se
- Rare Disease Sweden (one of several patient organizations), financed mainly by the state (the National Board of Health and Welfare) and project funding. www.sallsyntadiagnoser.se

Rare Disease Research Activities

Existence of rare disease research programmes/projects

There are currently no specific programmes or projects in Sweden to fund or facilitate rare disease research.

All expert teams that are participating in ERNs are actively involved in RD research (translational and clinical). Basic research covering different aspects of RD is carried out at most of the medical and natural science universities.

Participation in E-Rare and International Research Initiatives

Sweden participates in RD-ACTION, ICORD and is in collaboration with RDI through ICORD.

National Alliance of Patient Organisations and Patient Representation

There is a national alliance for rare disease patient organisations in Sweden. The work of Rare Diseases Sweden during the last year has resulted in the establishment of Centres of Expertise at all university hospitals. However the work has not resulted in a coordinating body, in order to completely meet the needs of the patient.

Rare Disease Sweden mainly work on a national level. Resources are limited yet Sweden has managed to have patient networks in close connection to the Centres of Expertise.

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.

Information and training for patients and families: Ågrenska Foundation family programs, www.agrenska.se.
Information for patients and families: The Swedish National Agency for rare diseases, www.nfsd.se

Information and training for caregivers: See section 11 and 13

Information for patients and families: Educational and information events are organised regularly and in collaboration with expert teams and RD patient organisations at the Karolinska Centre for Rare Diseases.

Rare diseases Sweden consists of national associations for a large number of various rare diseases, that on a voluntarily, not paid, basis give mutual support to its members.

Individual care plans are available to patients which provide access to social and support services. According to the Swedish legislation individual care programmes and permanent health care contact should be provided when there is a need of coordination.

Future plans include further training for medical professionals and an integrated medical and social (holistic) guidelines.

Orphan Medicinal Products

According to the Medical Products Agency (MPA) out of the 78 orphan medical products (OMPs) authorised by December 2012.

The Dental and Pharmaceutical Benefits Agency had decided to reimburse 38 OMPs.

Rare Disease Day

Rare diseases Sweden has arranged minor activities at the Rare Disease Day in 2014 and 2015, by arrangements in the Swedish Parliament. In 2016, there were two large events: A rare Film Festival and a seminar. These events, in 2016, resulted in a lot of publicity.

The so-called political week in Almedalen, Visby, Sweden, has taken place for more than 40 years. During this event politicians and influential people gather to debate current issues. More than 35.000 people participate during the event in 2015.

In order to raise awareness about RD and the ongoing work in the RD area, the Swedish National Agency for rare diseases, Ågrenska and Rare Disease Sweden has joined together during the week in Almedalen every year since 2013.

Rare Disease Sweden strives to let its members participate in order to let them come into public view. The organisation has created a portrait exhibition with pictures of some of the member, with short version of their life stories on the back of each picture. We use this exhibition in many a varied context.

In 2015, several members of the organisation participated in Almedalen, by performing at speaker's corner on a square. The portrait exhibition was also shown there.

Other

By the beginning of 2017 there will be a Centre for Rare diseases on each of the university hospital sites in the country. From the point of view of Rare Diseases Sweden, there is a very real need for a national plan of action for rare diseases to be put in place, which should have a comprehensive approach, bring together and coordinate all the various activities for rare diseases, according to the needs of the patients.