2013 REPORT ON THE STATE OF THE ART OF RARE DISEASE ACTIVITIES IN EUROPE

STATE OF THE ART OF RARE DISEASE ACTIVITIES IN SWEDEN

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More information on the European Union Committee of Experts on Rare Diseases can be found at www.eucerd.eu.

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ACRONYMS

CAT - Committee for Advanced Therapies at EMA
CHMP - Committee for Medicinal Products for Human Use at EMA
COMP - Committee on Orphan Medicinal Products at EMA
DG - Directorate General
DG Enterprise - European Commission Directorate General Enterprise and Industry
DG Research - European Commission Directorate General Research
DG Sanco - European Commission Directorate General Health and Consumers
EC - European Commission
ECRD - European Conference on Rare Diseases
EEA - European Economic Area
EMA - European Medicines Agency
ERN - European reference network
EU - European Union
EUCERD - European Union Committee of Experts on Rare Diseases
EUROCAT - European surveillance of congenital anomalies
EUROPLAN - European Project for Rare Diseases National Plans Development
EURORDIS - European Organisation for Rare Diseases
FDA - US Food and Drug Administration
HLG - High Level Group for Health Services and Medical Care
HTA - Health Technology Assessment
IRDiRC – International Rare Diseases Research Consortium
JA - Joint Action
MA - Market Authorisation
MoH - Ministry of Health
MS - Member State
NBS - New born screening
NCA - National Competent Authorities
NHS - National Health System
PDCO - Paediatric Committee at EMA
RDTF - EC Rare Disease Task Force
WG - Working Group
WHO - World Health Organization
GENERAL INTRODUCTION

This document was produced by the Scientific Secretariat of the European Union Committee of Experts on Rare Diseases (EUCERD), through the EUCERD Joint Action: Working for Rare Diseases (N° 2011 22 01), which covers a three year period (March 2012 – February 2015).

The present report aims to provide an informative and descriptive overview of rare disease activities at European Union (EU) and Member State (MS) level in the field of rare diseases and orphan medicinal products up to the end of 2012. A range of stakeholders in each Member State/country have been consulted during the elaboration of the report, which has been validated as an accurate representation of activities at national level, to the best of their knowledge, by the Member State/country representatives of the European Union Committee of Experts on Rare Diseases. The reader, however, should bear in mind that the information provided is not exhaustive and is not an official position of the European Commission, its Agencies or national health authorities.

The report is split into six parts:

Part I: Overview of rare disease activities in Europe
Part II: Key developments in the field of rare diseases in 2012
Part III: European Commission activities in the field of rare diseases
Part IV: European Medicines Agency activities and other European activities in the field of rare diseases
Part V: Activities in EU Member States and other European countries in the field of rare diseases
Part VI: Activities at National level in each EU Member State and other European countries in the field of rare diseases

Each part contains the following description of the methodology, sources and validation process of the entire report, and concludes with a selected bibliography and list of persons having contributed to the report.

The present document contains the information from Parts II and V of the report concerning Sweden. A list of contributors to the report and selected sources are in annex of this document. For more information about the elaboration and validation procedure for the report, please refer to the general introduction of the main report.
RARE DISEASE ACTIVITIES IN SWEDEN

Definition of a rare disease
The Swedish definition of a rare disease is a disorder resulting in an extensive disability with a prevalence of no more than 1 in 10,000 inhabitants. The Swedish Medical Products Agency applies the European Regulation on Orphan Medicinal Products definition, a prevalence of no more than 5 in 10,000.

National plan for rare diseases and related actions
In June 2010, the National Board of Health and Welfare presented a report concerning the organisation of national resources for rare diseases to the Ministry of Health and Social Affairs.

In June 2010, the Swedish Government decided to establish a national focal point for coordination in the field of rare diseases, a €300,000 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. On 24 July 2010, the National Board of Health and Welfare was commissioned by the Swedish Government to establish this national focal point in the field of rare diseases.

In November 2010, the Swedish National Conference on Rare Diseases was held in Stockholm, to discuss a future national plan or strategy for rare diseases under the EUROPLAN project. The conference allowed stakeholders to meet to discuss a range of policy topics and helped put rare diseases on the national agenda, stimulating discussion concerning a national plan for rare diseases.

At the end of 2011 National Board of Health and Welfare announced the new National Function for Rare Diseases (NFSD - Nationella Funktionen för Sällsynta Diagnoser). Their work will include the promoting of coherence and coordination of health care resources for people with rare diseases and to accomplish increased coordination with the social insurance, employment services, social services, NGOs and other actors. They will 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 is 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 November 2012, the Swedish national strategy for rare diseases was transmitted to the government. For the moment the financial implications have not been considered.

On 26 November 2012, a Europlan national conference was held in Stockholm bringing together 140 stakeholders, to follow the elaboration of the national plan for rare diseases. The conference was organised by the Rare Diseases Sweden and focused on how to transform the strategy proposed by the National Board of Health and Welfare into a concrete plan of action to improve care for rare diseases. Health care providers, policy makers and user representatives from across the country signed up to take part in the discussion about how to put the strategy into action.

Centres of expertise
Sweden’s health care system is decentralised and run by 21 county councils/regions. In accordance with a 1990 agreement with the Federation of County Councils, the National Board of Health and Welfare has issued a catalogue of providers of specialist care, which is intended to provide recommendations on reference points for local administrators. These centres providing expertise are mostly located at university hospitals. The new NFSD will collect information concerning centres of expertise in Sweden. Some university hospitals receive funding to develop regional centres for rare diseases. Criteria for expert centres are under development.

Registries
There is a National Patient Registry funded by the National Board of Health and Welfare, including the International Classification of Diseases, Tenth Revision (ICD 10) based diagnoses for all in-patient and some out-

1 http://www.regeringen.se/sb/d/13214/a/148634
patient visits (including day surgery and specialist psychiatric care) from both private and public health care providers. This registry is mainly used for statistics.

The centres of expertise, run by county councils/regions, have developed local quality registries to allow them to monitor activities and results. Currently there are approximately 20 registries for various rare diseases. During 2012 a working group was established for the purpose of developing national registries for rare diseases which will provide a tool to measure and monitor relevant patient care activities for patients with rare diseases.

At the national level, around 70 National Quality Registries have been established and are supported by the Swedish Association of Local Authorities and Regions (SALAR). All National Quality Registries contain individual-based data concerning diagnosis, treatment interventions and outcomes. These registries are primarily general and do not solely concern rare diseases, although patients with rare diseases may be included, as in, for example, the Swedish Dementia Registry. SALAR encourages managers of registries to apply for funding to become a National Quality Register in order to increase quality of health care on a national level as well as the accessibility of the registry.

The Swedish Association of the Pharmaceutical Industry runs a pilot project that aims to improve the documentation of orphan medicinal products in clinical use through quality registries. At the moment, this project covers Chronic Myelogenous Leukemia, Idiopathic Thrombocytopenic Purpura and Pulmonary Arterial Hypertension.

Sweden contributes to the EUROCASE CF, EUROCAT, SCNIR, and AIR European registries.

Neonatal screening policy
For many years, a newborn screening programme has been in place for phenylketonuria, hypothyroidism, congenital adrenal hyperplasia, biotinidase deficiency and galactosemia in Sweden. However, since November 2010, the programme has been extended to twenty-four disorders. The required blood sample volume remains the same. The additional disorders screened are 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, glutaric aciduria type I and 2, beta-ketothiolase deficiency, citrullinemia, argininosuccinate lyase deficiency (ASA), arginase deficiency, maple syrup urine disease (MSUD), tyrosinemia type 1, propionic acidemia and homocystinuria.

Genetic testing
Genetic testing is mainly performed by the six clinical genetics units at the University Hospitals of Lund, Gothenburg, Linköping, Uppsala, Stockholm and Umeå. Some molecular testing, mainly SNP-analysis for single polymorphisms, is done in medical biochemistry units or pathology units without special competence in clinical genetics. There are neither national reference laboratories nor any formal agreements between laboratories regarding co-operation and specialisation. There are no specific national guidelines for genetic testing issued by health authorities.

Genetic tests are reimbursed in the same way as all other medical tests. Laboratories must be authorised by the county councils in order to receive reimbursement (this applies for any laboratory service). There is no private sector of any significance. Genetic testing abroad is possible and is widely used, and there are not any specific regulations opposing this.

Diagnostic tests are registered as available in Sweden for 119 genes and an estimated 175 diseases in the Orphanet database.

National alliances of patient organisations and patient representation
The Swedish national patient organisation for rare diseases (Riksförbundet Sällsynta diagnoser) is a national alliance for rare disease patient organisations. The alliance aims to create a holistic view of the common problems associated with rare diagnoses to support small handicap groups, to ease the particular difficulties of patients with rare diagnoses and to promote and protect human rights. Riksförbundet Sällsynta Diagnoser also provides funds to support the empowerment of patient organisations.

Patient organisations for rare diseases are mainly sponsored by private sponsors, but they may also receive public sponsorship for specific projects. Although the Swedish healthcare system emphasises both decentralisation and organisation at municipal, county and regional levels, new bills have been passed by the
Parliament to support patient organisations and their activities. One bill supported a conference in November 2009 for members and non-members of Riksförbundet Sällsynta Diagnoser. Another bill supports further development of a communication platform on the website www.sallsyntadiagnoser.se, where a diagnosis database for members will be available. This communication platform will also allow those concerned to reach both formal and non-formal contacts and get in contact directly via a web community and a web forum.

At present, there is no specific platform for the representation of or consultation with patient organisations in policy issues for rare diseases in Sweden.

Sources of information on rare diseases and national help lines

Orphanet activities

Since 2006 the Karolinska Institute has hosted an Orphanet team in Sweden. During 2012 the Orphanet team was integrated with the Regional Centre of Rare Diseases, Karolinska University Hospital. The team collects data on services in Sweden related to rare diseases (for example, specialised clinics, medical laboratories with lists of diagnostics tests, ongoing research, registries, clinical trials, networks, technological platforms, patient organisations and emergency guidelines) for entry into the Orphanet international database. Since 2011, the team provides national information on the Orphanet Sweden website6, offering a national entry point to the Orphanet database in Swedish, giving medical professionals, patients, researchers and other interested parties free access to an encyclopedia and inventory of more than 3000 rare diseases, disease related gene description, orphan medical products and specialised services in Sweden and in 35 other countries. All data are reviewed by experts and abstracts are available in five languages. Orphanet Sweden started a close cooperation during 2012 with the recently installed National Function Point for Rare Diseases and the Information Centre of Rare Diseases to optimise the national information resources. In 2011 and 2012 Orphanet arranged conferences related to the Rare Diseases Day.

A nationwide survey was carried out at the end of 2010 to investigate the primary care of patients with rare diseases. The survey was based on the results of previous rare disease patient surveys and included questions concerning the types of rare diseases encountered by doctors, the diagnostic tools currently in use, what problems doctors face in the care of rare disease patients and how these can be solved. A second aim of the survey was to inform doctors about existing diagnostic tools and Orphanet services. The results demonstrate the importance and need of improving the education of rare diseases for GPs as well as informing professionals about existing web portals for rare diseases, such as Orphanet.

Official Information Centre for Rare Diseases

Since 1999, the Swedish Rare Disease Database has been run by the Swedish Information Centre for Rare Diseases (Informationscentrum för ovanliga diagnoser) at the Sahlgrenska Academy of the University of Gothenburg. The Centre is financed by the National Board of Health and Welfare and is a national resource for patients, families and professionals. Apart from producing and maintaining the Rare Disease Database, the Centre offers assistance in information retrieval and works to increase awareness and knowledge about rare diseases. The Centre acts as a clearing house for all information related to rare diseases and for relevant national resources.

The Rare Disease Database of the National Board of Health and Welfare currently includes detailed and expert-validated information about 300 rare diseases. This material is freely accessible at the website of the National Board of Health and Welfare: www.socialstyrelsen.se/ovanligadiagnoser. Each text in the database includes information on symptoms, occurrence, causes, genetics, diagnostics, treatment, national and regional resources, patient associations, courses for patients and for healthcare professionals, national medical specialists, national (and sometimes international) medical centres, social support, other sources of information, and research references.

The material can be printed out easily. There are also concise information sheets for every disease. All the texts in the database are updated on a regular basis. The material is being translated into English and currently more than 160 information texts can be accessed at www.socialstyrelsen.se/en/rarediseases. The database has more than 1 million visitors per year.

Help line

The Swedish Information Centre for Rare Diseases also serves as a public helpline by answering questions, mediating contacts and giving advice on where to find further assistance. Orphanet Sweden also provides information about national and international rare diseases resources.

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6 http://www.orphanet.se/national/SE-SV/index/hemsida/
Other sources of information on rare diseases

Ågrenska contributes to the dissemination of information by providing information with a holistic perspective, including information on available social services. They also participate in maintaining the Nordic web resource www.rarelink.org and the Ågrenska Academy was established in 2009 and provides streaming live cast lectures and conferences. Experiences and lecturers are documented and distributed on Ågrenska’s website (about 150 documents).

There is a close cooperation between Ågrenska and the Swedish National Organisation for Rare Diseases.

Updated information on orphan medicinal products has been published by Läkemedelsindustriföreningen (LIF), the trade association for research-based pharmaceutical industry in Sweden.

Riksförbundet Sällsynta Diagnoser has an online database with information on 60 rare diseases.

Good practice guidelines

Professional networks between the University Hospitals of Sweden and within Medical Associations initiates and continuously develops best practices and guidelines for rare diseases. The Best practice clinical guidelines have been elaborated upon as a result of initiatives by Ågrenska and professional networks.

Training and education initiatives

A number of courses are held on the initiative of patient organisations and knowledge centres for rare diseases. Ågrenska offers families, adults and children the possibility to benefit from educational activities adapted to their needs. They also provide guidance regarding the availability of social services. In 2012 Ågrenska arranged twenty-five national family stays such as empowerment programmes, including two educational days for professionals and six empowerment programmes for adults (adult stays) with a rare disease.

The University hospital teams that provide care for certain rare diseases educate and inform patients and families during educational days about their specific diagnosis. Medical professionals and representatives from the social services are given specialist training. The Orphanet team also helps specialists in training about how to find validated information on rare diseases.

National rare disease events in 2012

Rare Diseases Sweden organised a number of events to mark Rare Disease Day. The campaign kicked off in the months prior to the Day and included a photography exhibition and a lecture tour over the winter of 2011-2012 to establish contact between the Swedish Alliance of Rare Diseases and hospitals working with rare disease patients.

On 29 February 2012, volunteers organised an event at the Central station of Stockholm to reach the general public and raise awareness. On the same day, a number of meetings and think tanks were held: a range of stakeholders were invited to discuss the content of the national plan/strategy of rare diseases.

A rare disease scholarship was bestowed to a young volunteer in a rare disease patient organisation to mark the day. In addition a flash mob was organised to mark the Day.

On 22 February a regional meeting to celebrate the Day organised by Orphanet in Stockholm together with the Swedish Alliance of Rare Diseases under the auspices of the Karolinska University Hospital and the Karolinska Institutet. The theme of the meeting was “Shaping regional centres for rare diseases” and was attended by about hundred representatives of health care professionals, researchers, patients, pharma industry, health authorities and politicians. The regional meeting was one of a series of Regional meetings across Sweden to mark Rare Disease Day.

On 26 November 2012, a Europlan conference was held in Stockholm, to follow the elaboration of the national plan for rare diseases.

Rare Diseases Sweden held their annual meeting on 24-25 November 2012, welcoming 50 participants.

Hosted rare diseases events in 2012

No specific reported activity.

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7 http://www.lif.se/
Hosted Nordic Events
The Nordic Council has funded a project that investigated possible areas for Nordic networking and cross-country cooperation in the field of rare diseases. One of these projects is arranging Nordic conferences.

Research activities and E-Rare partnership

National research activities
The Swedish Research Council (SRC) is a government agency under the Ministry of Education and Science. The agency evaluates and prioritises research in medicine, pharmacy, odontology and dental care sciences and decides on project grants in these fields. Project funding is based on quality criteria (bottom-up procedure) and not subject to prioritisation based on research area, with a few exceptions. SRC also makes decisions to provide financing for principal investigators in areas of research where directed support is of strategic value. Rare diseases are thus funded through a yearly call for proposals for project grants; however, there is no dedicated budget for rare diseases. Instead, applications dealing with rare diseases compete with other applications on the basis of the quality of the proposal and not subject to prioritisation of research areas, with a few exceptions. Approximately 80 research projects on rare diseases were funded by SRC.

Medical research on rare diseases is also supported by a number of private foundations. However, these grants are not specifically dedicated to rare diseases.

Research on rare diseases is performed at many universities and university hospitals. This research is supported by grants from the government as well as from non-governmental foundations. Clinical research concerning rare diseases is partly supported by county councils/regions and clinical trials are partly sponsored by orphan drug companies. Some 50 national hospital units and 30 university departments involved in research activities are registered in the Orphanet database.

The Swedish Cancer Society and the Childhood Cancer Foundation are examples of a non-profit organisation which contributes to the funding of cancer research (including rare cancer), information-sharing and supporting activities which aim to improve cancer treatment and care. Research projects are funded following the same policy as that of the SRC.

It is impossible to separate support for rare disease research from support for orphan medicinal product development, as these research efforts are often mixed. In all likelihood, however, probably very little money directly supports orphan drug development.

An example of a centre performing research on rare disorders is Mun-H-Centre. Their activities focus on oral health and orofacial functions such as eating, speech, facial expression and saliva control in rare diseases. Since 1996, data on oral health and orofacial function have been collected through structured parental and clinical observations and registered in a database. Selected data from the database is presented at the Mun-H-Centre website and the information is updated regularly.

The Family programme and Respite service at Ågrenska provides the opportunity to meet a large number of children with rare diseases. During family stays using an assessment form (validated by University of Gothenburg, Institute of Psychology), Ågrenska performs systematic observations of the children in their school, pre-school and leisure activities, and the results are put together in a database.

National participation in European research projects
Swedish teams participate or have participated in the following European research projects for rare diseases: AIPGENE, ACADEMIC GMP, ANTEPRION, BIOMALPAR, BNE, CHD PLATFORM, CUREHLH, CLINIGENE, DRUGSFORD, DSMLIFE, EMVDA, EUMITOCOMBAT, ENCCA, EURAPS, EUCLYD, EUROSD, EUROBONET, EUROGENETEST, EUROPEAN LEUKEMIA NET, EVI-GENORET, EMSA-SG, EUROCRA, EURADRENAL, EURAMY, EURO-GENE-SCAN, GENESKIN, FNAIT, HDLMOICS, IMPACTT, INTREALL, INNOVALIV, INHERITANCE, NMD-CHIP, LYMHPANGIOGENOMICS, MANASP, MOLDIAG-PACA, NEUPROCF, NEOTIM NEUROPRION, NEWTBDRUGS, PRIBOMAL, PWS, TRYPOBASE, TB-DRUG OLIGOCOLOR, TREAT-NMD, RD PLATFORM, RDCONNECT, RAREBESTPRACTICES, and VITAL.

E-Rare
Sweden is not currently a partner of the E-Rare project.

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6 Inventory of Community and Member States’ incentive measures to aid the research, marketing, development and availability of orphan medicinal products (2005 revision) (pp20-21).

9 www.mun-h-center.se
**IRDiRC**
Swedish funding agencies have not yet committed funding to the IRDiRC. However, the progress of the consortium activity programme is followed by Orphanet Sweden and the National Board of Health and Welfare.

**Orphan medicinal products**

**Orphan medicinal product committee**
A few Orphan medical product expert committees in Sweden have been formed on the initiative of the Swedish Society of Medicine and of local county councils, respectively.

**Orphan medicinal products incentives**
The Medical Products Agency (MPA)\(^{10}\) is responsible for the regulation and surveillance of the development, manufacturing and marketing of medicinal products in Sweden. Regarding orphan medicinal products, centralised marketing authorization application to the European Medicines Agency (EMA) is mandatory. The applications are assessed by the Committee for Human Medicinal Products (CHMP) at the EMA and the decision, valid for the whole of the EU, is granted by the Commission.

The MPA can waive the fees for clinical trial applications and provide scientific advice for researchers, applicants and companies lacking support from the pharmaceutical industry. Concerning the provision of free of charge IMP by clinical trial sponsors, Swedish law allows exemptions: should an obligation to perform a trial after marketing have been a condition of the marketing authorisation being granted for an orphan drug. The same could apply for all clinical trials and IMPs, not just orphan drugs on the condition that the clinical trial is performed without the participation of the pharmaceutical industry or that the clinical trial is special importance to public health.

**Orphan medicinal product market availability situation**
According to the MPA, out of the 78 orphan medical products (OMPs) authorised by December 2012, The Dental and Pharmaceutical Benefits Agency had decided to reimburse the following 38 OMPs (some with restrictions): Fabrazyme\(^*,\) Replagal\(^*,\) Glivec\(^**(for CML), *(withdrawn from registry of OMPs for remaining orphan designated conditions), Tracleer\(^**,\) Somavert, Zavesca (for Niemann-Pick’s disease), Carbaglu, Aldurazyme, Ventavis, Onsenal (withdrawn register medicinal products human use), Wilzin, Xagrid, Orfadin, Prialt, Revatio, Nexavar, Sutent\(^*,\) Thelin (withdrawn register medicinal products human use), Exjade, Sprycel, Diacomit, Inovelon, Cystadane, Revlimid, In creelex, Tasisina, Thalidomide Celgene, Volibris, Firazyr, Ceplene, Mepact, Afinitor\(^*,\) Cayston, Nplate, Revolade\(^*,\) Esbriet, TOBI Podhaler, Votubia. The DPBB decided not to reimburse the following 21 OMPs: Busilvex, Lita k, Lysodren, Pedea, Xyrem\(^*,\) Naglazyme, Myozyme, Evoltra, Savene, Elaprase, Soliris, Atiriance, Gilullan, Yon delis, Torisel, Kuvan, Vidaza, Mozobil, Ilaris\(^*,\) Arzerra, VPRIV\(^{11}\). Of note, Zavesca\(^**\) for treatment of Gaucher’s disease has been removed from the list of reimbursed OMPs by the DPBB. Reimbursement information is lacking for a number of OMPs, some of which have been withdrawn from the market but the majority being products authorised late in 2011 and 2012. Several OMPs are marketed in Sweden without general reimbursement.

**Orphan medicinal product pricing policy**
The Dental and Pharmaceutical Benefits Agency whether a new medicinal product should be reimbursed for community use and approves its pharmacy purchase and selling price. Manufacturers of hospital-use drugs can negotiate directly with the county councils\(^{12}\).

**Orphan medicinal product reimbursement policy**
Reimbursement decisions are made by The Dental and Pharmaceutical Benefits Agency\(^{13}\), a government agency commissioned to make decisions on state subsidies for dental and pharmaceutical products. If a positive decision on reimbursement has been made by the agency, OMPs are fully reimbursed by social insurance in Sweden (there are no conditions specific to orphan drugs) and are available through hospital and community pharmacies when prescribed by a specialist physician or a general practitioner.

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\(^{10}\) www.mpa.se

\(^{11}\) *The product has been withdrawn by the sponsor from the Registry of Orphan Medical Products, **The product has been removed from the Registry of Orphan Medicinal Products since its 10 years of market exclusivity has expired. For Glivec the applies to the acute lymphatic leukaemia indication only.

\(^{12}\) Orphan Drugs in Europe: Pricing, Reimbursement, Funding and Market Access Issues, Donald Macarthur (2011) p117

\(^{13}\) www.tlv.se
**Other initiatives to improve access to orphan medicinal products**
Compassionate use of OMPs/non-OMPs has been introduced in Sweden under the responsibility of the MPA. For OMPs not yet available in Sweden, the MPA can approve “named patient prescription” of a certain drug for a certain patient on a yearly basis: this procedure also applies for non-OMPs.

**Other therapies for rare diseases**
No specific information reported.

**Orphan devices**
No specific information reported.

**Specialised social services**
Ågrenska offers families, adults and children the possibility to benefit from programmes that provide guidance to patients regarding latest medical knowledge, available social services as well as educational and holistic activities adapted to their needs.

The Mo Gård Group coordinates measures for patients with communication disabilities, some of which are linked to rare diseases.

The Swedish Act concerning Support and Service for Persons with Certain Functional Impairments (*Lagen om stöd och service till vissa funktionshindrade* - LSS) is an entitlement law that ensures good living conditions for people with extensive and permanent functional impairment, ensuring that they receive the help they need in daily life and that they can influence the support and services they receive. This law is most relevant for rare diseases because in most cases, rare diseases entail functional impairment. Accordingly, municipality institutions provide fully reimbursed activities, such as respite care services, therapeutic recreational programmes and services aimed to promote the quality of life.  

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**DEVELOPMENT OF RARE DISEASE ACTIVITIES IN 2012 IN SWEDEN**

**National plan for rare diseases and related actions**
In November 2012, the Swedish national strategy for rare diseases was transmitted to the government. For the moment the financial implications have not been considered.

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**Centres of expertise**
The new NFSD will collect information concerning centres of expertise in Sweden. Criteria for expert centres are under development.

**Registries**
During 2012 a working group was established for the purpose of developing national registries for rare diseases which will provide a tool to measure and monitor relevant patient care activities for patients with rare diseases.

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14 [http://www.vgregion.se/upload/HoH/Kansli/H%c3%a5d%20och%20st%c3%b6d/lss-engelska-hso-hoh.pdf](http://www.vgregion.se/upload/HoH/Kansli/H%c3%a5d%20och%20st%c3%b6d/lss-engelska-hso-hoh.pdf)
Sources of information on rare diseases and national help lines

Orphanet activities
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The MPA can waive the fees for clinical trial applications and provide scientific advice for researchers, applicants and companies lacking support from the pharmaceutical industry. Concerning the provision of free of charge IMP by clinical trial sponsors, Swedish law allows exemptions: should an obligation to perform a trial after marketing have been a condition of the marketing authorisation being granted for an orphan drug. The same could apply for all clinical trials and IMPs, not just orphan drugs on the condition that the clinical trial is performed without the participation of the pharmaceutical industry or that the clinical trial is special importance to public health.

Orphan medicinal product market availability situation
According to the MPA, out of the 78 orphan medical products (OMPs) authorised by December 2012 The Dental and Pharmaceutical Benefits Agency had decided to reimburse the following 38 OMPs (some with restrictions): Fabrazyme**, Replagal**, Glivec**(for CML), *(withdrawn from registry of OMPs for remaining orphan designated conditions), Tracleer**, Somavert, Zavesca (for Niemann-Pick’s disease), Carbaglu, Aldurazyme, Ventavis, Onsenal (withdrawn register medicinal products human use), Wilizin, Xagrid, Orfadin, Prialt, Revatio, Nexavar, Sutent*, Thelin (withdrawn registar medicinal products human use), Exjade, Sprycel, Dicormit, Inovelon, Cystadane, Revlimid, Increlex, Tasigna, Thalidomide Celgene, Volibris, Firazyr, Ceplene, Mepact, Afinitor*, Cayston, Nplate, Revolade*, Ebriet, TOBI Podhaler, Votubia. The DPBB decided not to reimburse the following 21 OMPs: Busilvex, Litak, Lysodren, Pedea, Xyrem*, Naglazyme, Myozyme, Evoltra, Savene, Elaprase, Soliris, Atriance, Glilolan, Yondelis, Torisel, Vidaza, Mozobil, Ilaris*, Arzerra, VPRIV*5. Of note, Zavesca**

15 www.mpa.se
16 *The product has been withdrawn by the sponsor from the Registry of Orphan Medical Products, **The product has been removed from the Registry of Orphan Medicinal Products since its 10 years of market exclusivity has expired. For Glivec applies to the acute lymphatic leukaemia indication only.
for treatment of Gaucher’s disease has been removed from the list of reimbursed OMPs by the DPBB. Reimbursement information is lacking for a number of OMPs, some of which have been withdrawn from the market but the majority being products authorised late in 2011 and 2012. Several OMPs are marketed in Sweden without general reimbursement.
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SELECTED BIBLIOGRAPHY AND SOURCES

- Swedish Information Centre for Rare Diseases http://www.socialstyrelsen.se/
- Ågrenska http://www.aggrensa.se/en/
- National Quality Register http://www.kvalitetsregister.se/
- National Function for Rare Diseases www.nfsd.se

17 The contributors and validators of the report have contributed information which is accurate to the best of their knowledge. However, readers should take note that the contents of this report are illustrative and not exhaustive.
18 All websites and documents were last accessed in May 2013.
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- Orphanet Sweden national website
  http://www.orphanet.se/national/SE-SV/index/hemsida/
- Rare Diseases Sweden
  http://www.sallsyntadiagnoser.se/
- Medical Products Agency
  www.mpa.se
- Rarelink Sweden
  www.rarelink.se