

2014 REPORT ON THE STATE OF THE ART OF RARE DISEASE ACTIVITIES IN EUROPE



STATE OF THE ART OF RARE DISEASE ACTIVITIES IN SWITZERLAND

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The European Union Committee of Experts on Rare Diseases (EUCERD) was established in 2009 and its mandate ended in 2013. It is replaced from 2014 by the Commission Expert Group on Rare Diseases. The EUCERD Joint Action continues to support the activities of the new Expert Group until 2015.

More information on the activities of the former European Union Committee of Experts on Rare Diseases and the EUCERD Joint Action 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 European Union Committee of Experts on Rare Diseases (EUCERD) was established in 2009 and its mandate ended in 2013. It is replaced from 2014 by the Commission Expert Group on Rare Diseases. The EUCERD Joint Action continues to support the activities of the new Expert Group until 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 2013. 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 Commission Expert Group 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 2013

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

Parts I – V also include a description of the methodology, sources and validation process of the entire report, and 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 Switzerland. 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.

Each year, there are around 15 000 downloads of the different sections of the report combined.

RARE DISEASE ACTIVITIES IN SWITZERLAND

Definition of a rare disease

The Therapeutic Products Act (TPA) adopted the European Regulation on Orphan Medicinal Products definition of a prevalence of no more than 5 in 10 000 individuals and this applies for the simplified authorisation of orphan medicinal products. Stakeholders in Switzerland accept the European Regulation on Orphan Medicinal Products definition of a prevalence of no more than 5 in 10 000 individuals.

National plan/strategies for rare diseases and related actions

There is still no national concerted plan or strategy for rare diseases in Switzerland.

With the aim of filling this gap, on 16 December 2010, Ruth Humbel, member of the Health Commission in the Parliament submitted to the National Council a postulate for "*a national strategy for improving the health situation of people with rare diseases*". The National Council followed the recommendation of the Federal Council and accepted the claim in March 2011. The Federal Council has consequently assigned the Federal Office of Public Health the task of submitting a proposal. An alliance¹ encompassing patient organisations, the Swiss Medical Association, university hospitals, the rare disease informational portal Orphanet-Switzerland and representatives from the pharmaceutical industry, then joined forces in August 2011 to promote a national strategy for rare diseases in Switzerland. Chaired by National Councillor Ruth Humbel, this newly-formed community of interest for rare diseases (*IG rare diseases*) is actively engaged in advocating the development of a national action plan for the country's rare disease patients. In September 2012, National Councillor Guy Parmelin requested information concerning the state of advancement of the national strategy for rare diseases. The answer of the Federal Council included the information that a formal meeting between the IG and the Federal Office of Public Health took place on 4 June 2012 to define the expectations and priorities for the development of a national plan. Issues such as financing diagnostic tests and defining the criteria for centres of expertise have been broached. On Rare Disease Day 2013 Pascal Strupler, Director of the Federal Office of Public Health confirmed that the elaboration of a national concept for rare diseases would take place in the second quarter of 2014². In 2013 the Federal Office of Public Health recruited a "Rare diseases project manager" to oversee the assessment of the situation of rare diseases in Switzerland and bring stakeholders together. A number of stakeholder meetings were organised in 2013 to advance with the elaboration of the concept. In addition a working group of the Swiss Academy of Science worked on the definition of rare diseases and proposed a set of criteria when establishing reference centres in Switzerland. The concept focuses on issues such as the difficulties to pose an adequate diagnosis in a timely fashion, the provision of high quality medical care, mechanisms that strengthen the resources available to the patients and their relatives as well as to support research projects at national and international level. The Swiss Conference of the Cantonal Ministers of Public Health (GDK/CDS) also supports the publication of a manual for the employees of the information and counselling centres for prenatal testing and coordinates the offer of highly specialised medicine in Switzerland, including rare disease patients. The only specific project for rare diseases supported by the GDK/CDS is Orphanet Switzerland.

Centres of expertise

Several specialised care centres have been established as centres of reference by reputation, usually in University Hospitals. In addition to this, the Inter-Cantonal Agreement on Highly Specialised Medical Services (IAHSMS) coordinated by the GDK/CDS came into effect in 2009 the purpose of which is that "*the cantons shall agree, in the interests of a needs-based, high-quality and economical health care system, to ensure coordination in relation to the centralisation of highly specialised medical services. This applies to those medical fields and services that are characterised by their rarity, by their high potential for innovation, by high personnel or technical costs or by complex treatment procedures. For categorisation as a highly specialised medical service, at least three of the aforementioned criteria must be met, whereby rarity must always apply*". The appointed centres can be consequently considered as official reference centres of expertise³. In 2011, several centres have been officially appointed in the fields of metabolic diseases, retinoblastoma, primary immunodeficiency in children, surgery of the liver and biliary tract in children, rare medullar tumours, surgery of epilepsy and

¹ http://www.orphanet.ch/PDF/MEDIENMITTEILUNG_Gruendung_IG_Seltene_Krankheiten_f_def.pdf

² <http://www.orphanet.ch/PDF/ProRaris25022013cloture.pdf>

³ <http://www.gdk-cds.ch/index.php?id=903&L=1>

neurosurgery of complex vascular anomalies of the central nervous system. In 2013, new centres have been named in the field of paediatric oncology (haematopoietic stem cells transplants (autologous and allogeneic), treatment of neuroblastoma, treatment of soft tissue sarcomas and other bone tumours and central nervous system tumours), adult oncology (haematopoietic stem cells transplants) and cochlear implants.

Registries

There are a number of registries for specific rare diseases in Switzerland. Switzerland contributes/contributed to the following European registries: AIR, CAPS, E-IMD, E-HOD, TREAT-NMD, EURO CARE-CF and EUROCAT.

Neonatal screening policy

A newborn screening programme covering all of Switzerland is in place and includes screening for phenylketonuria, congenital hypothyroidism, galactosaemia, congenital adrenal hypoplasia, biotinidase deficiency, and medium-chain acyl-CoA dehydrogenase (MCAD) deficiency. The request to implement the neonatal screening for cystic fibrosis, initiated by the Swiss Cystic Fibrosis Task Force, was approved from the Federal Office of Public Health for a 2-year-pilot project starting in January 2011, and extended with an open end date from 2013.

Genetic testing

The medical genetics speciality exists for laboratory directors (FAMH) and for medical doctors (FMH) and several specialised care and/or testing centres have been established as centres of reference by reputation, usually in University Hospitals. Genetic testing laboratories require formal authorisation⁴ to practice from the government; more than 60 public and private laboratories provide genetic testing, although not all tests are reimbursed. Since 2011, interlaboratory comparisons (EQA or other) must be performed at least once per year for every analysis proposed by genetic testing laboratories. Genetic counselling is formally required and is usually provided by doctors specialised in medical genetics or by referring doctors.

The efforts of genetic health professionals led to the approval on 2 December 2010 by the Federal Department of Home Affairs of the introduction (as of 1 April 2011) of an orphan disease regulation for the reimbursement of genetic laboratory testing of rare genetic diseases by the compulsory health insurance even if this test did *not* appear in previous list of approved tests or if the test is carried out abroad. An individual application for reimbursement is required and has to be submitted to the health insurance medical examiner (HIME) responsible.

Diagnostic tests are registered as available in Switzerland for 396 genes and an estimated 472 diseases in the Orphanet database⁵; this information, however, is not yet complete and does not cover all of Switzerland.

National alliances of patient organisations

Since 2004, Orphanet Switzerland has identified about one hundred rare disease patient organisations, some of them being related to international networks. Since 2009, the “strategic” position of Orphanet Switzerland with regards to contacts with patient organisations, has actively contributed to the creation of an Alliance of Rare Diseases in Switzerland, facing the challenges of uniting patients from four different linguistic areas. ProRaris, the Swiss Rare Disease Patient Alliance, was founded on 26 June 2010 representing 42 patient organisations.

In 2011, ProRaris⁶, the then newly founded Alliance, put all its efforts into increasing community awareness of rare diseases in Switzerland. In the framework of the third International Rare Disease Day in Switzerland in February 2013, a conference jointly organized by ProRaris and the newly founded Radiz (Rare Disease Initiative Zurich) took place at the University of Zurich. The conference topic was “putting together our know-how to increase the quality of care for rare diseases”. Stakeholders of various fields (politics, insurance, administration, medicine etc.) were present and discussed issues related to the formation of a national strategy for rare diseases (see also the announcement of the Director of the Federal Office of Public Health, Mr. Pascal Strupler above). The event received excellent press coverage, including articles in widely read papers in all regions of Switzerland, as well as radio and TV-interviews.

At the end of August 2012 ProRaris submitted a case study on the unequal treatment of the reimbursement of drugs for patients with rare diseases among various insurance companies in different

⁴ Federal Act on Human Genetic Testing (HGTA) of 8 October 2004, <http://www.admin.ch/ch/e/rs/8/810.12.en.pdf>

⁵ Information extracted from the Orphanet database (January 2014).

cantons to the director of the Federal Office of Public Health, Mr. Pascal Strupler. In his answer he announced a close monitoring of the interpretation of articles 71 a and b of the ordinance on health insurance (Krankenversicherungsverordnung) on which the decisions of the insurance companies are based until the end of 2013. An evaluation was carried out in 2013⁷.

As a patient representative, ProRaris is part of the “Community of Interest for rare diseases” and is strongly implied in the elaboration of the national concept for rare diseases.

Sources of information on rare diseases and national help lines

Orphanet activity in Switzerland

Since 2001 there is a dedicated Orphanet team in Switzerland, currently hosted by the Genetics and Laboratory Medicine Department of the University Hospital of Geneva. This team is composed of a country coordinator and, since 2011 of 2 information scientists (1 full time position and 1 part time position). Orphanet Switzerland has a close collaboration with the Health On The Net foundation (HON) for the management of the online forms. The team is in charge of identifying sources of information, collecting and updating data on rare disease related services (specialised clinics, medical laboratories, ongoing research, registries, clinical trials and patient organisations) at national level for entry into the Orphanet database. In 2011 the team launched the Orphanet Switzerland national website⁸ and contributes to the dissemination of information regarding the Orphanet database tool and national initiatives in the field of rare diseases (publications, media coverage, Orphanews, conferences, booth at annual congresses of learned societies, etc.). The website is currently only available in French. Additional resources are necessary to translate the content in German and Italian.

As a collaborating partner of the Orphanet Joint Action, Orphanet Switzerland is not entitled to the EU funding and must ensure its funding at national level. In 2011, the Swiss Conference of the Cantonal Ministers of Public Health (GDK/CDS) guaranteed a global budget for the period 2011-2014 for Orphanet.

Official information centre for rare diseases

There is no official information centre for rare diseases, however Orphanet is the reference portal for information on rare diseases and orphan medicinal products in Switzerland.

Help line

There is currently no help line available for rare diseases in Switzerland. However, a joint venture between the University Hospital of Lausanne (CHUV) and the University Hospital of Geneva (HUG) with the support of Orphanet Switzerland was initiated in 2012 in order to create a regional portal on rare diseases and to establish a helpline which was launched in 2013 (www.infomaladiesrares.ch). This francophone portal provides access to current information on specialised clinics and allows a greater visibility of local and regional resources, based on existing Orphanet data. This service will be completed by a Helpline in 2014.

Other sources of information on rare diseases

The GDK/CDS supports the establishment of cantonal information and counselling centres for prenatal testing and the HGTA requires non directive genetic counselling before and after genetic testing. However, the existence of four national languages complicates the organisation of collective national projects. Orphanet Switzerland is currently the only rare disease project supported financially by the GDK/CDS.

The Federal Office of Public health publishes the list of the laboratories with an authorisation to execute genetic tests.⁹

In 2013 the “Romand (French speaking part of Switzerland) portal on rare diseases” (www.info-maladies-rares.ch) was launched.

Guidelines

No specific information reported.

Training and education initiatives

On 4 -6 July 2013 Radiz (Rare Diseases Initiative Zürich), a clinical research priority program of the University of Zurich, organised its first yearly summer school on rare diseases at the lake of Zurich. The summer school’s main goal is to motivate bright young clinicians and scientists to work in the field of rare disease and to make

⁷ Evaluation of the implementation of article 71 a and 71 b KVV, accessible at <http://www.bag.admin.ch/evaluation/01759/02074/13897/index.html?lang=de>

⁸ <http://www.orphanet.ch>

⁹ <http://www.bag.admin.ch/themen/medizin/00683/02724/03677/index.html?lang=de>

them aware of the many opportunities, but also the challenges. The summer school focuses on a wide variety of subjects in the rare disease arena, from disease mechanisms and animal models, to improving diagnoses and novel therapeutics, with lectures and workshops on drug development, model organisms, how to choose clinical endpoints, clinical trials, regulatory aspects, patient registries, patient initiated research, ethical considerations, as well as what rare diseases may tell us about common diseases. The 2013 summer school was attended by 30 young clinicians and researchers. A 2nd Radiz summer school will take place in July 2014.

National rare disease events in 2013

To mark Rare Disease Day 2013, a number of events were organised and initiatives launched. On 22 February the University of Zurich, University Children's Hospital Zurich and University Hospital Zurich joined forces to launch Radiz with a kick-off meeting followed by a networking dinner with the Gebert-Rüf foundation and a press release entitled "An end to the Odyssey"¹⁰. The following day (23 February 2013) ProRaris, in partnership with Radiz, organised for the 3rd year in a row a conference to discuss the situation for rare disease patients in Switzerland at the University of Zurich which was attended by stakeholders and key political decision makers in Switzerland. The Day was also marked by the launch of the "Romand (French speaking part of Switzerland) portal on rare diseases" www.info-maladies-rares.ch.

Hosted rare disease events in 2013

Amongst the rare disease events hosted in Switzerland in 2013 and announced in *OrphaNews* was Autoinflammation 2013 - 7th International Congress of the International Society of Systemic Auto-Inflammatory Diseases (22- 26 May 2013, Lausanne).

Research activities and E-Rare partnership

National research activities

Although there is no specific national budget for rare disease research, the Telethon Suisse raises funds for rare diseases, specifically for neuromuscular disorders. Moreover, many projects on rare diseases are supported by the Swiss National Science Foundation and a few public foundations (i.e. the Gebert Rüf Foundation and the BLACKSWAN Foundation). A new clinical research priority program of the University of Zurich - Radiz - was started in the fall of 2012. Radiz is funding translational research projects and supporting and training young clinicians and researchers with the aim to increase awareness for rare diseases and stimulate interdisciplinary collaborations.

The *Gebert Rüf Foundation*¹¹, a Swiss grant programme specifically for rare diseases, announced its fifth call for projects in 2013. The independent foundation is committing CHF2 million (€1.66 million) per year to researchers based at Swiss universities, university hospitals, federal institutes of technology and universities of applied sciences. The Rare Diseases – New Approaches grant programme, which launched in 2009, is established as a five-year area of activity. The initiative aims at developing and implementing innovative technologies or approaches in the diagnosis and treatment of rare diseases. In 2013 5 finalists were chosen from the 73 submissions received. The knowledge gained should lead to a better understanding of the genetic, molecular and biochemical processes underlying these diseases and pave the way towards new forms of treatment or diagnostics. A further aim is to improve the transfer of basic research findings into clinical practice. The focus must be on innovation, feasibility and effectiveness, while attaining high scientific and technological standards.

The *BLACKSWAN Foundation*⁶ is active since 2009 and supports advanced research into rare diseases in order to complement the chronic lack of public and private funds in this area. The principal goals are to promote and fund therapeutic application of new scientific protocols in order to find effective treatments and to increase public understanding and awareness of rare diseases.

Participation in European research projects

Swiss teams participate/have participated in 55 rare disease related FP7 projects and 2 projects were coordinated by a Swiss team.

¹⁰ www.orphanet.ch/PDF/press_release_Radiz.pdf

¹¹ <http://www.grstiftung.ch/en.html> & http://www.grstiftung.ch/en/portfolio/activity/active_areas/rare-diseases.html

⁶ <http://www.blackswanfoundation.ch/>

E-Rare

Since 2013 Switzerland is a member of the E-Rare project and the BLACKSWAN Foundation is an active collaborator of the network. Switzerland participated in the 5th Joint Transnational call in 2013 with Swiss teams participating in 2 out of the 12 consortia selected for funding.

IRDIRC

Swiss funding agencies have not yet committed funding to the International Rare Diseases Research Consortium (IRDIRC).

Orphan medicinal products

The Swiss Orphan Drug Regulation was introduced in 2006: this regulation stipulates that orphan medicinal product status applies to products treating diseases affecting no more than 1 in 2000 persons. The availability of orphan medicinal products has been improved since 2006 thanks to the simplified authorisation procedures and the recognition of the orphan medicinal product status for any drug for which this status has been granted in a country with a comparable drugs authority.

Orphan medicinal product committee

No specific activity reported.

Orphan medicinal product incentives

Companies acquiring orphan medicinal product designation for their products are allowed tax exemption for certain administrative taxes but are not however allowed market exclusivity.

Orphan medicinal product market availability situation

140 active ingredients for 225 indications subject to the orphan drugs regulation are marketed in Switzerland⁸.

Orphan medicinal product pricing policy

Compared to European Member States the pricing and reimbursement procedure in Switzerland is considered relatively quick and is speeded up when drugs target unmet medical needs or show high therapeutic benefit¹².

Orphan medicinal product reimbursement policy

On 23 November 2010 the Federal Supreme Court decided that a health insurer was not obliged to reimburse the treatment costs (500'000 Swiss Francs per year) of Myozyme[®] for a patient with Pompe Disease, on the grounds that the therapy costs are not proportionate to the expected benefits for this specific patient. Based on this case, the Federal Court of Justice decided to fix limits for reimbursements, and although regretting the risk of unfairness, admits that rationing must be introduced. A limit of 100,000 Swiss Francs (€83'000) per year per patient has been proposed.

As of 2 February 2011 the Federal Council put two new articles of the Federal Ordinance on the Health Insurance into force stipulating that the off label use of drugs and the treatment with drugs not listed on the list of the reimbursed drugs (Spezialitätenliste) is admitted in case of life-threatening diseases if an important therapeutic benefit is expected from the treatment and if there is no reimbursed alternative. The Ordinance gives the insurers the freedom to decide about the maximum amount to be reimbursed.

Other initiatives to improve access to orphan medicinal products

No specific activity reported.

Other therapies for rare diseases

No specific activity reported.

Orphan devices

No specific activity reported.

⁷ Orphan Drugs in Europe : Pricing, Reimbursement, Funding and Market Access Issues, Donald Macarthur (2011) p120

⁸ <http://www.swissmedic.ch/daten/00081/index.html?lang=de>

Specialised social services

No specific activity reported.

DEVELOPMENT OF RARE DISEASE ACTIVITIES IN 2013 IN SWITZERLAND

National plan/strategies for rare diseases and related actions

On Rare Disease Day 2013 Pascal Strupler, Director of the Federal Office of Public Health confirmed that the elaboration of a national concept for rare diseases would take place in the second quarter of 2014¹³. In 2013 the Federal Office of Public Health recruited a “Rare diseases project manager” to oversee the assessment of the situation of rare diseases in Switzerland and bring stakeholders together. A number of stakeholder meetings were organised in 2013 to advance with the elaboration of the concept. In addition a working group of the Swiss Academy of Science worked on the definition of rare diseases and proposed a set of criteria when establishing reference centres in Switzerland. The concept focuses on issues such as the difficulties to pose an adequate diagnosis in a timely fashion, the provision of high quality medical care, mechanisms that strengthen the resources available to the patients and their relatives as well as to support research projects at national and international level.

Centres of expertise

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Neonatal screening policy

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¹³ <http://www.orphanet.ch/PDF/ProRaris25022013cloture.pdf>

¹⁴ Evaluation of the implementation of article 71 a and 71 b KVV, accessible at <http://www.bag.admin.ch/evaluation/01759/02074/13897/index.html?lang=de>

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Research activities and E-Rare partnership

National research activities

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¹⁵ <http://www.bag.admin.ch/themen/medizin/00683/02724/03677/index.html?lang=de>

¹⁶ www.orphanet.ch/PDF/press_release_Radiz.pdf

¹⁷ <http://www.grstiftung.ch/en.html> - http://www.grstiftung.ch/en/portfolio/activity/active_areas/rare-diseases.html

⁶ <http://www.blackswanfoundation.ch/>

applied sciences. The Rare Diseases – New Approaches grant programme, which launched in 2009, is established as a five-year area of activity. The initiative aims at developing and implementing innovative technologies or approaches in the diagnosis and treatment of rare diseases. In 2013 5 finalists were chosen from the 73 submissions received. The knowledge gained should lead to a better understanding of the genetic, molecular and biochemical processes underlying these diseases and pave the way towards new forms of treatment or diagnostics. A further aim is to improve the transfer of basic research findings into clinical practice. The focus must be on innovation, feasibility and effectiveness, while attaining high scientific and technological standards.

E-Rare

Since 2013 Switzerland is a member of the E-Rare project and the BLACKSWAN Foundation is an active collaborator of the network. Switzerland participated in the 5th Joint Transnational call in 2013 with Swiss teams participating in 2 out of the 12 consortia selected for funding.

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¹⁸ 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.

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¹⁹ All websites and documents were last accessed in May 2014.