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2012 REPORT ON THE STATE OF THE ART OF RARE DISEASE ACTIVITIES IN EUROPE OF THE EUROPEAN UNION COMMITTEE OF EXPERTS ON RARE DISEASES

STATE OF THE ART OF RARE DISEASE ACTIVITIES IN SWITZERLAND

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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

General

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

Pilot European Reference Networks

Dyscerne - European network of centres of expertise for dysmorphology
ECORN-CF - European centres of reference network for cystic fibrosis
Paediatric Hodgkin Lymphoma Network - Europe-wide organisation of quality controlled treatment
NEUROPED - European network of reference for rare paediatric neurological diseases
EUROHISTIONET - A reference network for Langerhans cell histiocytosis and associated syndrome in EU)
TAG - Together Against Genodermatoses – improving healthcare and social support for patients and families affected by severe genodermatoses
PAAIR - Patients' Association and Alpha-1 International Registry Network
EPNET - European Porphyria Network - providing better healthcare for patients and their families
EN-RBD -European Network of Rare Bleeding Disorders
CARE-NMD -Dissemination and Implementation of the Standards of Care for Duchenne Muscular Dystrophy in Europe project
ENERCA - European network for rare and congenital anaemia – Stage 3

GENERAL INTRODUCTION TO THE REPORT ON THE STATE OF THE ART OF RARE DISEASE ACTIVITIES IN EUROPE OF THE EUROPEAN UNION COMMITTEE OF EXPERTS ON RARE DISEASES

The 2012 Report on the State of the Art of Rare Disease Activities in Europe 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 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 2011. 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 either the European Commission, its Agencies or national health authorities.

The report is split into five parts:

Part I: Overview of rare disease activities in Europe
Part II: Key developments in the field of rare diseases in 2011
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

Each part contains a 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 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[1].

[1] http://www.eucerd.eu/upload/file/Reports/2012ReportStateofArtRDActivities.pdf

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 alliance[2] 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.

The Federal Office of Public Health is working on a project that will facilitate the reimbursement of rare disease medicinal products. A round table meeting held on 23 September 2011 gave health professionals, representatives of the biopharmaceutical industry, health insurances, patient organisations and local government representatives the opportunity to exchange views. Amongst the topics broached were strategies for reimbursing products and evaluating their benefits, as well as ways to improve diagnosis, for which the French model of identifying and creating networks of expertise was evoked. Finally, the issue of negotiating prices for rare disease treatments was discussed, as well as the necessity for clinicians and researchers to collaborate to enhance the understanding of rare disease treatments. A second round table was held in early 2012. The project should then be open for consultation later in 2012.

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 expertise[3]. 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 neurosurgery of complex vascular anomalies of the central nervous system.

Pilot European Reference Networks

Switzerland has participated and participates in the following European Reference Networks for rare diseases: Dyscerne, E-IMD, ENERCA, EPI/EPNET and PAAIR.

[2] http://www.orphanet.ch/PDF/MEDIENMITTEILUNG_Gruendung_IG_Seltene_Krankheiten_f_def.pdf
[3] http://www.gdk-cds.ch/index.php?id=903&L=1

Registries

There are a number of registries for specific rare diseases in Switzerland. Switzerland contributes to the following European registries: AIR, CAPS, E-IMD, TREAT-NMD, EUROCARE-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 in December 2010 and a 2-year-pilot project started in January 2011.

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 authorisation[4] 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 296 genes and an estimated 320 diseases in the Orphanet database[5]; 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, ProRaris[6], as a newly founded Alliance, put all its efforts in the increasing of awareness of rare diseases in Switzerland. In the framework of the 4th International Rare Disease Day, ProRaris organised the first conference on rare diseases in Switzerland addressing the main topic "*Inequal access to health care*" with the lack of coverage by health insurances of genetic testing and orphan medicinal products

In addition to this major event, large media coverage has been achieved including a special television documentary on rare diseases and on the non-reimbursement for orphan products deemed too expensive (cf. Federal Court decision of 23 November 2010). The TV program[7] was followed by a live debate on rationing health costs with, among others, the participation of the director of the Federal Office of Public Health.

As a patients' representative, ProRaris is part of the *Community of Interest for rare diseases*, founded in August 2011, and is strongly implied in political advocacy for the elaboration of a national plan for rare diseases.

Within the framework of the new project supported by the European Commission to support rare disease national plans, the proposal of ProRaris to organize, by 2013, a EUROPLAN conference in Switzerland with all stakeholders, which will be supported by EURORDIS, was selected.

[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 (September 2011).
[6] www.proraris.ch
[7] « Too expensive: you die ! »: http://www.tsr.ch/emissions/36-9/3264987-trop-cher-tu-meurs.html

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) in their country for entry into the Orphanet database. In 2011 the team launched the Orphanet Switzerland national website[8] 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.).

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 president of the Swiss Conference of the Cantonal Ministers of Public Health (GDK/CDS) guaranteed a global budget for 2011 and 2012 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.

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]

Good practice guidelines

No specific information reported.

Training and education initiatives

No specific information reported.

National rare disease events in 2011

To mark Rare Disease Day 2011, the first conference on rare diseases in Switzerland was organised on 19 February 2011 in Bern by ProRaris, the new Swiss Alliance, and by Orphanet Switzerland. Around 450 participants, including patients, health professionals, scientists and politicians, met to learn, share and discuss the rare diseases public health issue.

Telethon Switzerland is a regular, annual fixture and organised a fund raising event at the start of December 2011 for rare diseases.

Hosted rare disease events in 2011

A number of rare disease related events were organised in Switzerland in 2011: Treat-NMD Global Conference (8-11 November 2011, Geneva), Society for the Study of Inborn Errors of Metabolism – Annual Symposium 2011 (30 August – 02 September 2011, Geneva), 2nd Annual World Orphan Drug Congress (29 November – 1 December 2011, Geneva), Sanfilippo Foundation Switzerland International Congress (8-10 December 2011, Geneva).

[8] http://www.orpha-net.ch/?lng=FR

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

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).

The *Gebert Rüf Foundation*[10], a Swiss grant programme specifically for rare diseases, announced its third call for projects in 2011. 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. The first two calls in 2009 and 2010 selected ten finalists from 106 applications. In 2009, the chosen topics were: Preventing Nodule Formation in Hyaline Fibromatosis Patients; Genetic Screening for Disease-Causing Mutations in Familial Polycythemia Using Next Generation DNA Sequencing; Gene Hunting for Recessive Hereditary Peripheral Neuropathies by Recent and Highly-Parallel Technologies; Hereditary Sensory Neuropathy Type 1 - Pathomechanism and Therapy; and Identification of New Factors Implicated in Genetic Gonadal Disorders. In 2010, the chosen topics were: Towards a better mechanistic understanding of Friedreich's Ataxia; Role of macroautophagy in CGD and correction of the defect; Consanguinity and rare recessive disorders; Rescue of dysfunctional RNA processing in spinal muscular atrophy through PGC-1-alpha; and Novel mechanisms causing Lafora disease.

In 2011, the chosen topics were: Prodrug Platform for Rare Colonic Diseases; Treatment for Dysferlinopathies; Vaccination for the Prevention and Cure of Inflammatory Bowel Disease; Host- and Pathogen-Derived Factors in Chronic Mucocutaneous Candidadiasis; Rational Targeting of FOXC2 Haploinsufficiency; and Role of snoRNAs in the Development of Prader Willi Syndrome. 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

Switzerland participates or has participated in European rare disease research projects including: AAVEYE, ANTIMAL, AUTOROME, BIOMALPAR, CLINIGENE, CSI-LTB, CSI-LTB, E-IMD EMVDA, EURADRENAL, EURO-LAMINOPATHIES, EUGINDAT, EURAPS, EUREGENE, EUROBONET, EUROGENTEST, EUROGLYCANET, EUROPEAN LEUKEMIA NET, EVI-GENORET, GENESKIN, GEN2PHEN, HDLOMICS, HUMALAB, IMMUNOPRION, LEISHMED, LYMPHANGIOGENOMICS, MYELINET, MILD-TB, MPCM, MYORES, NEUROPRION, NANOTRYP, NOVSEC-TB, NM4TB, PEMPHIGUS, PULMOTENSION, TRYPOBASE, THERAPEUSKIN, and SIOPEN-R-NET.

E-Rare

Switzerland is not currently a member of the E-Rare project although the BLACKSWAN Foundation is an active collaborator of the network.

IRDiRC

Swiss funding agencies are not currently committed members of 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

[10] http://www.grstiftung.ch/en.html
[6] http://www.blackswanfoundation.ch/

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
At least 36 orphan medicinal products with EU market authorisation are marketed in Switzerland[8].

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 benefit[11].

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.

Orphan devices
No specific activity reported.

Specialised social services
No specific activity reported.

DEVELOPMENT OF RARE DISEASE ACTIVITIES IN 2011 IN SWITZERLAND

National plan/strategies for rare diseases and related actions
With the aim of filling this gap in rare disease policy in Switzerland, 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

[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

Council has consequently assigned the Federal Office of Public Health the task of submitting a proposal. An alliance[12] 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.

The Federal Office of Public Health is working on a project that will facilitate the reimbursement of rare disease medicinal products. A round table meeting held on 23 September 2011 gave health professionals, representatives of the biopharmaceutical industry, health insurances, patient organisations and local government representatives the opportunity to exchange views. Amongst the topics broached were strategies for reimbursing products and evaluating their benefits, as well as ways to improve diagnosis, for which the French model of identifying and creating networks of expertise was evoked. Finally, the issue of negotiating prices for rare disease treatments was discussed, as well as the necessity for clinicians and researchers to collaborate to enhance the understanding of rare disease treatments. A second round table was held in early 2012. The project should then be open for consultation later in 2012.

Centres of expertise

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 neurosurgery of complex vascular anomalies of the central nervous system.

Neonatal screening policy

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 in December 2010 and a 2-year-pilot project started in January 2011.

Genetic testing

Genetic testing laboratories require formal authorisation[13] 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.

National alliances of patient organisations

In 2011, ProRaris, as a newly founded Alliance, put all its efforts in the increasing of awareness of rare diseases in Switzerland. In the framework of the 4th International Rare Disease Day, ProRaris organised the first conference on rare diseases in Switzerland addressing the main topic "*Inequal access to health care*" with the lack of coverage by health insurances of genetic testing and orphan medicinal products.

As a patients' representative, ProRaris is part of the *Community of Interest for rare diseases*, founded in August 2011, and is strongly implied in political advocacy for the elaboration of a national plan for rare diseases. Within the framework of the new project supported by the European Commission to support rare disease national plans, the proposal of ProRaris to organise, by 2013, a EUROPLAN conference in Switzerland with all stakeholders, which will be supported by EURORDIS, was selected.

[12] http://www.orphanet.ch/PDF/MEDIENMITTEILUNG_Gruendung_IG_Seltene_Krankheiten_f_def.pdf
[13] Federal Act on Human Genetic Testing (HGTA) of 8 October 2004, http://www.admin.ch/ch/e/rs/8/810.12.en.pdf

Sources of information on rare diseases and national help lines

Orphanet activity in Switzerland

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 president of the Swiss Conference of the Cantonal Ministers of Public Health (GDK/CDS) guaranteed a global budget for 2011 and 2012 for Orphanet. In 2011 the team launched the Orphanet Switzerland national website[14].

National rare disease events in 2011

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Research activities and E-Rare partnership

National research activities

The *Gebert Rüf Foundation*[15], a Swiss grant programme specifically for rare diseases, announced its third call for projects in 2011. In 2011, the chosen topics were: Prodrug Platform for Rare Colonic Diseases; Treatment for Dysferlinopathies; Vaccination for the Prevention and Cure of Inflammatory Bowel Disease; Host- and Pathogen-Derived Factors in Chronic Mucocutaneous Candidadiasis; Rational Targeting of FOXC2 Haploinsufficiency; and Role of snoRNAs in the Development of Prader Willi Syndrome. 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.

IRDiRC

Swiss funding agencies are not currently committed members of the IRDiRC.

Orphan medicinal products

Orphan medicinal product reimbursement policy

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.

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[14] http://www.orpha-net.ch/?lng=FR
[15] http://www.grstiftung.ch/en.html
[6] http://www.blackswanfoundation.ch/

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- Black Swan Foundation
  http://www.blackswanfoundation.ch/
- Association Enfance et Maladies Orphelines
  http://www.aemo.ch/index.php?page=news/afficher&idNew=22

[16] 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.
[17] All websites and documents were last accessed in May 2012. A more detailed list of sources is available in the full report: http://www.eucerd.eu/upload/file/Reports/2012ReportStateofArtRDActivities.pdf