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

Lithuania Report

Definition of a rare disease

Lithuania has adopted the European Commission's definition of a rare disease. Stakeholders in Lithuania accept the European Regulation on Orphan Medicinal Products definition of a prevalence of no more than 5 in 10,000 individuals.

National Plan/Strategy

On 18 October 2012, the national plan for rare diseases was approved by Order No V-938 of the Minister of Health, and a National rare diseases coordination committee was formed, including delegated experts from university hospitals, universities, non-governmental organisations, state institutions representatives to oversee the plan.

The plan aims to establish a common approach on rare diseases, to raise public awareness, and to ensure prevention, early diagnosis, and efficient treatment, improvement of quality of life and social support for patients suffering from rare diseases. It also includes the optimisation of health care services and rational allocation of available resources, as well as measures for improving the assessment of medicinal products and medical devices.

The National plan for rare diseases has its list of measures for the period of 2013-2017, which is expected to be reviewed and developed for the period of 2018-2022.

A policy/decision has been made to ensure long-term sustainability of the measures contained within the National Plan. There is no specific budget for the National plan for rare diseases. Expenses for health care services and drugs for rare diseases patients are reimbursed from the Compulsory Health Insurance Fund budget as for other groups of patients. There are specific allocations for reimbursement of rare medicinal products and devices. Additionally, expenses for the treatment of ultra-rare diseases abroad are reimbursed from Compulsory Health Insurance Fund budget (Order No. 1566 of 30th December 2015 of the Minister of Health); compensation for orphan medicinal products and medicinal devices for ultra-rare diseases and conditions, unexpected cases are paid from a selected part of the budget of the Compulsory Health Insurance Fund (Order No. 1577 of 31st December 2015 of the Minister of Health). In 2016 4.7 million euros were allocated for the reimbursement of medicinal products and devices in the case of ultra-rare diseases (in 2015 3.19 million euros were allocated, but actual costs reached 4.36 million euros).

At the moment RDs cases are registered in clinical databases which are operated by university hospitals (Vilnius University Hospital Santaros klinikos and Hospital of Lithuanian University of Health Sciences Kauno klinikos). The established e. health system will have the statistical analysis subsystem for RD data collection and analysis. The next period (year 2016–2025) of Lithuania central eHealth system development will cover functionalities for specialized medicine areas, including functionalities for RD management.

The monitoring committee for the National plan for rare diseases was formed, including delegated experts from university hospitals, universities, non-governmental and patient organisations, state institutions representatives to oversee the plan implementation. Usually the national plan for rare diseases monitoring committee meets once per 6 months, in case of necessity – once per quarter.

The National Plan has had many successes to date:

- Centres of competence are established in university hospitals by the order of the head of the university hospitals. These university hospitals provide diagnostics, treatment, research, training and health care services by teams of health professionals based on multidisciplinary approach.
- A coordinating centre for diagnosis, treatment and management of rare diseases w established at the Vilnius University Hospital Santaros klinikos in November 2012.

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- Both Vilnius University Hospital Santaros klinikos and Hospital of Lithuanian University of Health Sciences Kauno klinikos has Coordinating Centres for rare diseases and centres of competence for different diseases are working under their supervision.
- A coordination centre for children with rare diseases was established at Vilnius Children's Hospital (Affiliate of Vilnius University Hospital Santaros klinikos).
- In order to strengthen the ERS rehabilitation services, there is also amended Children's Health Inspection Procedure (order No. V-383 of the Minister of Health of 23 March 2015). The regulations of mandatory preventive child health and young children (up to 4 years) psychomotor development testing requirements and procedures are approved by this order.
- Genetic testing is provided for patients of high risk group according to the order of the Minister of Health No. V-1458 of 31 December 2014; the expenses related to this testing are reimbursed from Compulsory Health Insurance Fund budget. Basic prices for reimbursement of genetic testing are approved by the same order of the Minister of Health (2014-12-31, No. V-1458).
- Genetic testing abroad is reimbursed on referral by the university hospital's consilium and decision made by Commission at the Ministry of Health (Order of the Minister of Health 2010-08-16 No. V-729).
- Genetic testing is available in two main university hospitals and several private laboratories. Only the clinical geneticist can order the genetic testing at academic hospitals, as established in order of the Minister of Health on 2012-08-02, No. V-745.
- Centre for Medical Genetics (CMG), Vilnius University Hospital Santaros klinikos (VUH SK). CMG is appointed as an institution performing mandatory nationwide neonatal screening for phenylketonuria, congenital hypothyroidism, galactosaemia and congenital adrenal hyperplasia according to the Orders of the Minister of Health (2014-05-22, No V-601; 2014-12-31, No V-1457; 2016-06-30, No V-873).
- Requirements of palliative care services for adults and children new edition adopted by the Order No. V-946 of the Minister of Health of 13 August 2015.
- In The Action Plan for reducing health inequalities in Lithuania, 2014-2023, approved by Order No V-815 of the Minister for Health of the Republic of Lithuania "On the approval of the Action Plan for reducing health inequalities in Lithuania, 2014-2023" of 16 July 2014 (part 7)-activities for improving quality and accessibility of health care services for children with rare diseases are foreseen.
- Centres of Expertise designation procedure (including Centres of Expertise for RD) is approved by the Lithuanian Government decree No 226 of 9 March 2016. Ministry of Health of the Republic of Lithuania has endorsed 18 healthcare providers (university clinics) for the purposes of participation in European Reference Networks.
- A training program for the improvement of rare diseases diagnostics for doctors has been initiated and a training cycle called "Rare diseases" has been introduced for medical students.

Organisation of Rare Disease Health and Social Care

Centres of Expertise

The Centres of Expertise designation procedure (including Centres of Expertise for RD) is approved by the Lithuanian Government decree No. 226 of 9 March 2016.National Policy for designating centres of expertise for RD fully implemented. Lithuania has approved Centres of Expertise designation procedure.

Health care institutions seeking to become a European reference network members are assessed at the national level in order to comply with national legislation on personal health care services in Lithuania. If an institution gets confirmation of the Ministry of Health of the Republic of Lithuania of compliance with the national legislation, it can apply to become a member of European reference networks. European Commission takes the final decision on a specific health care institution to become a member of specific European reference networks.

The National criteria used to designate a CE for RD is in accordance with the EUCERD criteria.

According to the decree 2016 No. 226 of Government of the Republic of Lithuania, health care institutions seeking to get approval of compliance to the national legislation of the Republic of Lithuania, to the Ministry of Health must provide the following:

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- a request form with attached documents confirming that health care institutions seeking to get approval of compliance to the national legislation of the Republic of Lithuania, have the right to provide personal care services related to rare diseases in the area where are seeking to become a member of the European reference networks ;

- a description of activities of the individual health care institutions seeking to get approval of compliance to the national legislation of the Republic of Lithuania, expertise, experience, scope of activities, health care specialists and qualifications, functions, distribution, use of information technology, scientific research and training activities implementation of personal health care services related in the area to rare diseases, seeking to become provision, which aims to become a member of the European reference networks.

University hospitals provide diagnostics, treatment, research, training and health care services by teams of health professionals based on multidisciplinary approach. Holistic approach is ensured partly, as much as this is within the competence of medical services, including rehabilitation. Steps are being made for better development of collaboration with social and educational services in order to fully implement holistic approach.

European Reference Networks (ERNs)

The Ministry of Health of Lithuania has nominated 12 HCPs who have submitted their applications for full membership of several ERNs.

At present 2 HCPs are participating as full members of 12 ERNs.

Members	**European Reference Networks (ERNs)**
Hospital of Lithuanian University of Health Sciences Kauno Klinikos	Endo-ERN ERN EYE ERN Skin ERN EURACAN
Vilnius University Hospital Santariškių Klinikos	ERKNet ERN EuroBloodNet ERN eUROGEN ERN ITHACA MetabERN ERN PaedCan ERN TRANSPLANT-CHILD ERN-RND

Rare Disease Registration

There are neither national nor regional rare disease registries in Lithuania but disease specific registries are available.

Nationwide disease codification according to the International Statistical Classification of Diseases and Related Health Problems, Tenth Revision, Australian Modification (ICD-10-AM/ACI/ACS) is regulated by the Order of the Minister of Health (2011-02-23, No V-164). Data on RD patients having codes in ICD-10-AM/ACI/ACS are collected in Statistics Lithuania.

Disease specific data bases are available. Part IV, article 27 of the National RD plan (Order of the Minister of Health 2012-10-18, No V-938) states that systemic approach should be implemented to the solutions of RD codification and data collection problems by incorporating electronic, platform-based registries during the period of implementation of E-Health project. Registries of separate diseases (RD included) will function as subsystems of National E-Health information system and will be integrated into common data network.

The next period (year 2016–2025) of Lithuania central eHealth system development will cover functionalities for specialized medicine areas, including functionalities for RD management.

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In Lithuania morbidity data is calculated from Compulsory Health Insurance Fund Information System (CHIF IS). This information system covers primary, out-patient and inpatient services paid by CHIF. CHIF IS covers about 99% of inpatient care, up to 100% of primary health care and about 90% of out-patient care services in the country. CHIF IS includes all diagnosis registered for the person in primary, out-patient or inpatient care (main diagnosis, complications and co-morbidities) coded using ICD-10-AM (Australian modification). Using CHIF IS it is possible to calculated morbidity by rare diseases if they could be recognised by ICD-10-AM code.

In Lithuania few registers having some information on rare diseases exit: Tuberculosis register, Cancer register, Causes of death register.

The Tuberculosis register covers all newly registered tuberculosis cases and relapses. Physicians should report by filling in the standard reporting forms. The Hospital of Infectious Diseases and Tuberculosis, Affiliate of Vilnius University Hospital Santaros klinikos is responsible for the register.

The National Cancer Institute is responsible for the Cancer register. The Cancer register exists for more than 30 years. The Register covers all reported cases of malignant neoplasms and some other neoplasms (including cases registered in death certificates only). Physicians are required to report cases by completing the standard reporting form.

The Cause of death register was founded in 2010. Up until 2010 the organisation, Statistics Lithuania, was responsible for collating cause of death statistics. Since 2010 this responsibility has been given to the Institute of Hygiene. The Register issues all medical death certificates in Lithuania and provides information on deaths of Lithuanian nationals living abroad. Multiple causes of death are coded since 2010.

In Lithuania, tertiary level complex rare disease diagnostics and treatment services are mostly concentrated in two university hospitals: Vilnius University Hospital Santaros klinikos (VUH SK) and Hospital of Lithuanian University of Health Sciences Kauno klinikos (HLUHS KK), therefore, competence centres for rare diseases and clinics of these two institutions have capabilities of leading patient registries for selected rare diseases approaching those of regional level.

Currently, VUH SK hosts about 40 institutional databases for rare diseases / groups of rare diseases patients including haematological diseases (extended registry including outcome measure indicators), congenital heart defects in children and adults, hearing impairments, rare epilepsies, rare kidney diseases, pulmonary hypertension, cardiomyopathies and others.

Additionally, teams of VUH SK contribute to European/international registries including ECARUCA, ESID, EuroCMR registry, EuroMyositis registry, EuroCMR registry, ROPAC, EACTS Database for Congenital Heart Disease Surgery, EUROFEVER, ARegPKD, ESPN/ERA-EDTA, KAPPA, PRINTO- PRES, GalNet, and E-HOD.

HLUHS KK hosts these rare diseases databases: Hypertrophic cardiomyopathies; rare renal diseases in children and adults (with DNA biobank); tuberous sclerosis (participation in international TOSCA registry); neurofibromatosis type I; neuromuscular disorders in children; epidermolysis bullosa; ichthyosis; autoimmune bullous dermatoses; purulent hidradenitis; monogenic diabetes type I (participation in international registry); hypopituitarism (participation in international registry); hypophysis adenoma; Turner syndrome; thyroid cancer; congenital adrenal hyperplasia; rare childhood tumours; neuroendocrine tumours; solid childhood tumours (in collaboration with Nordic Society of Paediatric Haematology and Oncology (NOPHO, under development); neurooncology; biological brain tissues; Fabry disease (incl. screening); genetic diseases

A nationwide registry and a biobank are available for diseases covered by nationwide neonatal screening at the Centre for Medical Genetics (CMG), Vilnius University Hospital Santaros klinikos (VUH SK). CMG is appointed as an institution performing mandatory nationwide neonatal screening for phenylketonuria, congenital hypothyroidism, galactosaemia and congenital adrenal hyperplasia according to the Orders of the Minister of Health (2014-05-22, No V-601; 2014-12-31, No V-1457; 2016-06-30, No V-873).

The Registry of ongoing clinical trials, including those for RD, is held by the State Medicines Control Agency at the Ministry of Health of the Republic of Lithuania, http://www.vvkt.lt/Informacija-apie-vykstancius-tyrimus

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Genetic Testing

Genetic testing is available in two main university hospitals and several private laboratories. Only the clinical geneticist can order the genetic testing at academic hospitals, as established in order of Minister of Health on 2012-08-02, No. V-745. Centre for Medical Genetics in Vilnius University Hospital Santaros klinikos and Centre for Rare diseases at the Hospital of Lithuanian University of Health Sciences Kauno klinikos provide genetics services and diagnostic services for rare diseases to the Lithuanian population.

Diagnostic tests are registered as available in Lithuania for 51 genes and an estimated 29 diseases in the Orphanet database. Different laboratories can perform several diagnostic tests which are registered in the local institutional database. Reference genetic testing laboratories do not exist in Lithuania. Different genetic testing laboratories apply adapted guidelines/protocols for Lithuania.

Centre for Medical Genetics (CMG), Vilnius University Hospital Santaros klinikos (VUH SK). CMG is appointed as an institution performing mandatory nationwide neonatal screening for phenylketonuria, congenital hypothyroidism, galactosaemia and congenital adrenal hyperplasia according to the Orders of the Minister of Health (2014-05-22, No V-601; 2014-12-31, No V-1457; 2016-06-30, No V-873).

Genetic testing is mostly provided as a part of clinical genetics services. Provision of genetic testing and clinical genetics services is regulated by the Order of the Minister of Health 2014-12-31, No V-1458 including indications for genetic counselling/clinical genetics services, ordering of genetic testing and reimbursement issues.

Clinical genetics services in VUH SK are carried out in compliance with international professional standards (including European Society of Human Genetics guidelines and recommendations) with pre- and post-testing genetic counselling. Additionally, psychologic, dietologic (mostly for patients with inborn errors of metabolism) and social services are provided, and multidisciplinary patient discussions by involvement of all required kind of medical specialists from VUH SK are regularly held.

Tests for the diagnostics of inborn errors of metabolism, molecular karyotyping and certain molecular genetic tests are provided in VUH SK only, while next-generation sequencing is provided in VUH SK mainly. Two other university hospitals, namely Hospital of Lithuanian University of Health Sciences Kauno klinikos (HLUHS KK) and Klaipeda University Hospital, provide some genetic counselling and genetic testing services, while genetic counselling services only provided in some other regional hospitals. Some genetic counselling and genetic testing services are also carried out in a private sector.

There is no official registry of clinical genetics/genetic testing services in Lithuania. However, a summary and a list of genetic tests provided in university hospitals are available at www.santa.lt and www.kaunoklinikos.lt. National laboratories provide data to the Orphanet Lithuania team.

Genetic testing services are fully reimbursed when provided through clinical genetics services covered by Compulsory Health Insurance Fund. Expenses of genetic counselling/genetic testing services are reimbursed according to the Order of the Minister of Health 2014-12-31, No V-1458, and follows a related test grouping model according to complexity/costs of tests.

Genetic testing abroad is possible in cases when genetic tests are not available in Lithuania, procedure of approbation and reimbursement for genetic testing abroad is regulated by the Order of the Minister of Health 2010-08-16, No V-729, with the appointment of special Commission at the Ministry of Health following clinical discussion and decision made by at least three clinicians.

Neonatal Screening

Universal neonatal screening is carried out in Lithuania for 4 congenital metabolic diseases: hypothyroidism (started in 1993), phenylketonuria (started in 1975), galactosaemia (started in 2015) and adrenal hyperplasia (Adrenogenital syndrome) (started in 2015). Universal neonatal screening for congenital metabolic diseases approved by order No. V-865 of the Minister of Health of 6 December 2004 “Regarding the Approval of Universal Screening of New-borns for Inborn Metabolism Disorders Procedures” (further amendments of this order made due to promoting further neonatal screening for congenital metabolic diseases).

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The red reflex test of neonatal eyes is also performed, which checks for the inherent optical media opacity and other eye diseases that prevent the normal development of the visual system, early detection and treatment (started in 2012, Order No. V-470 of the Minister of Health of 28 May 2012).

In order to improve the early diagnosis and treatment of neonatal hearing, health care institutions that provide obstetric services have been equipped with medical devices for new-born hearing testing. Hence, in 2014 universal new-born hearing testing in Lithuania was introduced (Order No. V-612 of the Minister of Health of 11 June 2013).

In 2016 Lithuania has also started a universal new-born screening for critical congenital heart defects (Order No. V-824 of the Minister of Health of 2 July 2015).

The further expansion of the neonatal screening is under discussion.

Guidelines and Training Activities

Clinical Practice Guidelines (CPGs)

You can find the main Lithuanian Clinical Practice Guidelines for rare diseases on the Ministry of Health webpage: (https://sam.lrv.lt/lt/veiklos-sritys/diagnostikos-gydymometodikos- ir-rekomendacijos/diagnostikos-ir-gydymo-metodikos) (available only in national language). The following are available:

- Diagnosis and treatment of hereditary metabolic diseases during neonatal period;
- Diagnosis and treatment of Celiac disease;
- Diagnosis and treatment of Cystic fibrosis;
- Diagnosis and treatment of Turner syndrome;
- Diagnosis, treatment and monitoring of congenital heart disease;
- Diagnosis and treatment of hypopituitarism;
- Diagnosis and treatment of arterial aneurysms and arteriovenous malformations;
- Diagnosis and treatment of glioblastoma.
- Development, adaption and implementation of CPGs are regulated by the orders of the minister. (Orders of the Minister of Health: 2006-05-17, No V-395; 2014-12-02, No V-1248).

Training and Education

Teaching and training on rare diseases is coordinated by the Faculty of Medicine at Vilnius University and the Lithuanian University of Health Sciences. Highly specialized academics, in every field of medicine, provide an in-depth teaching programme.

The Faculty of Medicine at Vilnius University has 7 departments, 11 clinics and 2 institutes and every division has a very specific role within the educational process.

Medical students begin learning about rare diseases during their first year through the teaching of General and human genetics, rare diseases are used as examples in the teaching of biochemical processes of the human body and students are also taught to assess the occurrence or recurrence of rare disease. In general, more than 38 academic hours are spent studying disciplines connected with rare diseases in the first year of medical school. Examples on rare diseases are used in every field of basic human metabolic processes while studying biochemistry, physiology and pathology at the time of preclinical studies, in total around 9 academic hours per year. Clinical studies starts at the second semester of the 3rd year of medical school. Students are taught about rare diseases in every clinical department until they graduate. Orthopaedics, general and plastic surgery, neonatology and paediatrics are the fields presenting rare diseases for 3rd year medical students, and it takes around 15 academic hours per semester. Obstetrics, gastroenterology, neurology, infectious diseases and rehabilitation subjects include rare diseases in the 4th year of medical school and learning takes around 40 academic hours per year. In the 5th year of medical school students are taught about rare diseases in many subjects: gynaecology, otorhinolaryngology, urology, nephrology, rheumatology and psychiatrics, and it takes around 43 academic hours per year. Medical students study rare diseases in paediatrics and clinical genetics disciplines in the final 6th year of medical school, which is around 50 hours per semester. The percent of

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information about rare diseases comparing to any other diseases varies from 2 percent (e.g. gastroenterology) to 100 percent (e.g. clinical genetics) in different subjects. Summarizing, medical students are familiar enough with rare diseases at their graduation.

In addition, there are special training courses performed by academics of Vilnius University Faculty of Medicine for postgraduate doctors that want to widen their knowledge of rare diseases and clinical genetics. Four different courses are suggested, each lasts for 36 academic hours:

1) Genetic laboratory tests,

2) Current clinical genetics,

3) Prenatal diagnosis of congenital anomalies,

4) Rare disease topical issues, hereditary cardiovascular pathology.

The Lithuanian University of Health Sciences and Hospital of Lithuanian University of Health Sciences Kauno klinikos training/education offer programs for undergraduate and postgraduate medical students, which include topics on rare diseases and rare disease policy (but this is only at the postgraduate level). The number of credits and contact hours differ by the frequency and clinical relevance of certain rare diseases in different medical specialities. Rare disease topics included within the undergraduate curriculum differ to those of postgraduate courses for different specialists. The Educational e-Portal on the official website of Hospital of Lithuanian University of Health Sciences Kauno klinikos available at:

https://portalas.kaunoklinikos.lt/User/LogOn?ReturnUrl=%2F
http://mokymai.kaunoklinikos.lt/course/index.php?categoryid=31

Information Resources for Rare Diseases

Orphanet Activities

Lithuania has an operational Orphanet team which currently housed within Vilnius University Hospital Santaros klinikos. The Orphanet information produced is available in the Lithuanian national language although no printed items are available. There is no specific funding available to for the activities of the Orphanet team however the team has achieved since the beginning of 2014:

- The dissemination and advancement of Orphanet information through conferences, the Rare Disease Day national campaigns and local websites.
- The analysis and review of international and national documents related to Orphanet and rare diseases. Collection of information on Centres of Expertise, medical laboratories, research etc.
- Applying for the European Reference Centres.
- Estimation and explanation of the enquiries of Orphanet related questions of the professionals, groups and patients. Implementation of Orphacodes in Vilnius University Hospital Santaros klinikos information system.

National Helplines

There is currently no helpline in Lithuania dedicated to rare diseases.

In the period of the 2014–2020 European Union funds' investment in the health sector will be pursued by coordinating integrated actions in order to ensure equal access (to a maximum possible extent) to the target groups to high-quality public services of health promotion, disease prevention, early diagnostic and specialised health-care. Investments will also be made in the introduction and development of efficient models for the organisation, provision and management of health-care services and timely aid by supporting innovative and efficient solutions and technologies to enhance the level of health literacy, to provide consultations, medical treatment and to observe target groups and to maintain their quality of life.

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Two projects are in the process of establishing National Helplines for children with rare diseases which will be coordinated by two university hospitals (Vilnius University Hospital Santaros klinikos and Hospital of Lithuanian University of Health Sciences Kauno klinikos). It is hoped that the projects will promote and develop experimental research and public health actions, as well as to provide technical expertise and information on rare diseases and orphan drugs, for the prevention, treatment and surveillance of these diseases. They will also act as the first contact point for information and communication for patients suffering from one of several thousand rare diseases and their families and collaborate with the national organisations of patients suffering from rare diseases.

Official Information Centres

Lithuania has official and unofficial information centres for rare disease. Information on rare disease can be found via the following means:

- The Coordinating Centre of Children's Rare diseases is the at Vilnius Children's Hospital (Affiliate of Vilnius University Hospital Santaros klinikos) within Vilnius University Hospital who can be contacted during working hours by phone and email (retos.ligos@vuvl.lt).
- Rare disease information can also be obtained via the rare disease charity and support foundation webpage (http://rarediseases.w7.lt/).
- The VUH SK Centre for Medical Genetics has a coordinating centre for rare diseases. It can be reached during working hours via telephone and email (genetika@santa.lt) and provides patients with information on rare disease issues. Web page: http://www.santa.lt/index.php?option=com_content&view=category&id=135&Itemid=429
- The official website of Hospital of Lithuanian University of Health Sciences Kauno klinikos (http://www.kaunoklinikos.lt/kk/), also the staff of the Coordinating Centre for Rare Diseases (retosligos@kaunoklinikos.lt) or by direct telephone contact) provide information on the services provided for rare disease patients at different centres and departments of the Hospital, also other relevant issues regarding RD, like national policy regarding RD management, patient organizations, information about certain RD, etc. Educational e-portal (https://portalas.kaunoklinikos.lt/User/LogOn?ReturnUrl=%2F) also serves as a source of information about multiple rare diseases.
- Also, activities in the field of comprehensive information on certain rare diseases organized by the staff or in collaboration with patient organizations in the form schools or summer camps (e.g., Turner syndrome, metabolic bone diseases, Spina biffida, Down syndrome). Multiple educational materials created and published by the staff on certain rare disease topics do exist and is being distributed.
- Also The Ministry of Health of the Republic of Lithuania provides information on RD issues on its web page (http://sam.lrv.lt/lt/veiklossritys/asmens-sveikatos-prieziura/retos-ligos).

No additional funding is provided for Coordinating Centres for Rare Diseases which perform information dissemination among institutions, patients etc. They are financed from the main budget of the institution.

Rare Disease Research Activities

Existence of Rare Disease Research programmes and projects

Lithuania has specific programmes to fund and facilitate rare disease research. These projects are funded through the general research budget.

Research which focusses on the ongoing social or socio-economic effects of rare diseases has been conducted in Lithuania. Projects have included:

1. "Relationship between psychological distress, inflammatory agents and brain natriuretic peptide with prognosis of brain tumours";

2. "Evaluation of physical and psychological wellbeing in hypopituitarism since childhood after treatment with growth hormone";

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3. “Correlations among endocrine hypophyseal functioning, body structure, psychological functioning and quality of life in head trauma patients“;

4. “Anthropometry, metabolic and psychological status in young and middle-aged hypogonadal men”;

5. “Optimisation of diagnostics and course of rare skin diseases by evaluating clinical, quality of life and healthcare service data“.

Also university hospitals participate in international projects which focus on clinical research but these are not state funded.

Participation in E-Rare and International Research Initiatives

Lithuania participates in the following research initiatives:

- Orphanet - www.orpha.net - we have Lithuanian team for the implementation of the Orphanet activities at national level.
- Health programme RD Action – tool aims to facilitate discussions and collaboration between experts willing to join an ERN. Promoting the implementation of recommendations on policy, information and data for rare diseases.
- Lithuanian-Swiss program Scientific Research and Development: Genetic diabetes in Lithuania.
- International Alexander von Humbold program-financed project entitled “Gels mimicking antibodies in their selective recognition of different molecular forms of human growth hormone“.
- Projects in collaboration with the Max Bergman Centre for Biomaterials, Dresden and the Leibniz Institute of Polymer Research, Dresden:

a. “Molecular Imprinting of Human Growth Hormone Dimers“(2014).

b. “Molecular Imprinting of the Fragments of the Human Growth Hormone“(2015).

COST activities:

c. BM 1105. “GnRH deficiency: Elucidation of the Neuroendocrine control of Reproduction”.

- TD1103. “European Network for Hyperpolarization Physics and Methodology in NMR and MRI” International E-epilepsy project
- 3rd Health Programme “Joint Actions on Rare Cancer, JARC“

International registries:

- Tuberous sclerosis registry (TOSCA)
- Hypopituitarism (participation in international registry)
- Monogenic diabetes type I (participation in international registry)
- Nordic Society of Paediatric Haematology and Oncology (NOPHO, under development)
- Gaucher disease

Three centres by September 8, 2016, for specific RD have successfully passed the 1st step of ERN eligibility check: ERN-skin, Endo-ERN, EURACAN, another one is still waiting for the decision to join ERN-EYE network.

National Alliance of Patient Organisations and Patient Representation

National Alliance – Association for Children Rare Diseases was founded in 2015 by the initiative of parents’ community who faced the diagnosis of rare disease with their children. The main mission of the Association is to represent interests and needs of patients with rare diseases focusing on children/paediatric population. Also to collaborate with specialists, represent patients’ needs with Health Care Institutions, Ministry of Health, to provide consultations for patients and their families, organize educational lectures and seminars for patients and their relatives.

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Over 2014-2015, parents community implemented the following activities: actively communicating and collaborating between parents with the help of social networks, continuously organizing patients/relatives meetings, developed internet website for specific disease areas. First National Conference for patients with rare disease took place in June, 2016.

More information about National Alliance activity: www.facebook.com/retosligos.

The Association for Children Rare Diseases unites and represents interest for already established small groups of patients as tuberous sclerosis, genetic neuromuscular disease and oesophagus atresia as well as separate patients' interests who are facing rare disease. Association for Children Rare Disease waiting for approval to became a member of EURORDIS organization.

Integration of Rare Diseases to Social Policies and Services

Specific actions exist to enable real access for people with rare diseases to general social/disability programmes (i.e. training, guidelines for social workers etc.) within Lithuania.

Social integration of disabled people is organised by applying the principles of equal rights, equal opportunities, discrimination prevention, of guaranteeing self-sufficiency and freedom of choice, accessibility, and compensation for disability, decentralisation and de-stigmatisation.

Social integration of the disabled and the quality of their life in society are ensured through measures such as adaptation of housing, payment of target compensations, support for disabled students, provision of technical aids, and promotion of tolerance.

The projects of social rehabilitation services in the community and support of association activities, targeted at the integration of the disabled, are financed. When implementing the projects, accessibility of services to the disabled is developed, self-sufficiency of the disabled is enhanced, and the role of the non-governmental sector is strengthened.

The provisions of this Convention are implemented through the National Programme for Social Integration of the Disabled 2013–2019 and the plans of implementing measures.

State budget funds for the implementation of programmes and measures

Title of the programme, measure	Amount used in 2012, EUR thous.	Amount used in 2013, EUR thous.	Amount used in 2014, EUR thous.	Amount used in 2015, EUR thous.	Amount allocated in 2016, EUR
Programme "Social Integration of the Disabled"	72 46.3	72 41.1	73 82.73	79 38.8	9 151
1. Developing accessibility of services for the disabled, enhancing self-sufficiency of the disabled and promoting their employment opportunities	56 64.3	5 686.5	5 537.2	5 815.4	5 631
2. Improving information environment for the disabled	239	186.1	447.2	441.7	336
3. Improving accessibility to public environment and adapting housing and surroundings thereof for persons with disabilities	945.5 835.4	860.5 846	856.1	1 168.2	1 631
4. Providing financial support to the disabled studying in higher schools	496.3	514.4	536.6	507.8	523
5. Paying benefits amounting to 20 per cent of BSB to the disabled who raise children for payment of public utilities, electricity or telephone bills or purchase of fuel	11.3	8.1	5.6	5.7	9

Data of the Department of Disabled Affairs under Table 1 the Ministry of Social Security and Labour.

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In 2012, when implementing the programme measure “Developing accessibility of services for the disabled, enhancing self-sufficiency and promoting their employment opportunities”, 408 projects selected by tender were financed; in 2013, the respective number of financed projects was 420; in 2014 – 413; in 2015 – 428; in 2016, 359 projects are financed. Each year, 60 municipalities finance projects of social rehabilitation services for the disabled in the community. These projects are implemented by organisations working in the field of social integration of the disabled. Moreover, the projects of supporting the activities of associations of the disabled and projects for the development of mobility and self-sufficient living skills of people with physical disabilities selected by tender and implemented by umbrella associations of the disabled were financed. In 2012, when implementing social integration projects financed by tender, over 45 000 persons with disabilities, including 17 per cent of persons with severe forms of disability, received various services (benefits); in 2013 – over 41 000, including 16 per cent of persons with severe forms of disability; in 2014–2015 – over 37 000, including 15 per cent of persons with severe forms of disability. The decrease in beneficiaries in 2014–2015 could be observed due to a stricter control of project results, regulated recommendations how to register projects of the recipients of services, thus the implementing organisations planned and implemented projects with a smaller number of disabled persons. When implementing measure “Improving information environment for the disabled”, the services of the Lithuanian sign language interpreters were provided. In 2012, these services were provided to 1 842 deaf people; in 2013 – 954; in 2014 – 759; in 2015 –808.

In 2013, 10 projects of financing the publishing and circulation of periodical publications for the disabled were selected by tender and financed; in 2014 and 2015 – 8 projects were financed each year. The projects of financing the publishing and circulation of periodical publications for the disabled were implemented by associations of the disabled and public institutions.

When implementing the measure “Adapting housing for persons with disabilities”, housing was adapted for the disabled, buildings that were relevant to the disabled were discussed in the information system “Infostatyba” and approved in construction accomplishment commissions.

When implementing measure “Improving accessibility to public environment and adapting housing and surroundings thereof for persons with disabilities”, projects of housing adaptation were implemented in accordance with the Description of the Procedure for Financing Housing Adaptation for People with Disabilities 2013–2015. As of 1 January 2016 the new version of the Description of the Procedure for Housing Adaptation for People with Disabilities 2016–2018 has become effective. When implementing the measure, works of adaptation of housing for the disabled are carried out, information on housing adaptation is collected, accumulated and disseminated, and associations of the disabled are involved in the control and supervision of the implementation of housing adaptation.

Housing adaptation expenses are covered from state and municipal budgets in parts: not more than 70 per cent (not more than 60 per cent as of 1 January 2017) from the state budget and at least 30 per cent (at least 40 per cent as of 1 January 2017) from the municipal budget for persons with very severe and severe movement and self-service dysfunctions; not more than 50 per cent from the state budget and at least 50 per cent from the municipal budget for persons with moderate movement and self-service dysfunctions. In 2012, 251 units of housing were adapted for the needs of the disabled; in 2013 – 224 units of housing; in 2014 –236 units of housing; in 2015 – 310 units of housing; in 2016, 350 units of housing are planned to be adapted.

When implementing the measure “Providing financial aids to disabled students”, financial support was further provided to disabled students in 2016. Following the Description of the Procedure for the Provision of Financial Aids to the Disabled Studying in Higher Schools, the funds are allocated for meeting special needs of the disabled studying in higher schools (a monthly benefit in the amount of 0.5 basic social insurance pensions) and partially reimbursing for studies (a benefit in the amount of 3.2 basic social benefit once in a semester).

In 2012, financial support was provided to 1 033 disabled students of higher schools; in 2013, support was provided to 1 080 disabled students; in 2014 – 1 021; in 2015 – 1 006; in 2016, support has been provided to 930 disabled students. Provision of technical aids for residents

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The measure “Acquisition and provision of technical aids for disabled people” of the Programme for Social Integration of the Disabled is implemented each year. The purpose of the measure is to develop the system of provision of country’s residents with technical aids (TA) and to meet the needs of provision with TA. Disabled people are provided with TA in four ways: provided free of charge from ten territorial units of the Centre of Technical Aid for Disabled People under the Ministry of Social Security and Labour (hereinafter referred to as the “Centre”) (in Vilnius, Kaunas, Klaipėda, Šiauliai, Panevėžys, Alytus, Utena, Telšiai, Marijampolė and Tauragė) or from municipal institutions; provided with TA bought by the Centre having paid additionally; acquire TA with their own funds and receive partial reimbursement for acquisition expenses; provided with TA through companies with which the Centre has concluded agreements under which companies receive compensations. Indicators of provision of the disabled with TA Year Overall TA provision level (per cent) TA for adults with paid compensations (per cent) TA for children with paid compensations (per cent) Total number of TA recipients Funds allocated,

Indicators of provision of the disabled with TA

Year	Overall TA provision level (per cent)	TA for adults with paid compensations (per cent)	TA for children with paid compensations (per cent)	Total number of TA recipients	Funds allocated, EUR
2012	94.1	90.7	97.7	34 473	2 038.3
2013	75.4	77.4	74.3	32 716	2 038.3
2014	75.1	73.4	84.3	34 123	2 294.4
2015	83.4	86.2	74.2	34 966	2 141.1
2016*	80	74	75	36 500	2 220.0

* (planned)

Data of the Centre of Technical Aid for Disabled People under Table 2 the Ministry of Social Security and Labour

The Centre and its units constantly provide information to society, collect feedback on TA, and consult on various ways of TA provision. The Centre regularly updates information about TA provision on its website and carries out publicity works (disseminates booklets, publishes articles in regional press). While improving accessibility to information about the provision of people with TA, meetings with medical specialists from personal health care institutions and municipality employees have been organised.

Vocational Rehabilitation Programme

Seeking to restore or increase the capacity for work level of the disabled and their professional competence and ability to participate in the labour market, the Vocational Rehabilitation Programme is implemented and vocational rehabilitation services are provided.

Seeking that disabled persons who complete the Vocational Rehabilitation Programme and become employed retain their employment as long as possible and in order to grant more responsibility for persons who have completed the Vocational Rehabilitation Programme to institutions providing vocational rehabilitation services, the Vocational Rehabilitation Programme (covering the following services: evaluation of professional competences, vocational guidance and counselling, restoration of professional competences or development of new competences, assistance when finding employment) was supplemented with a new service in 2014 – support at the workplace. It is the provision of assistance to the employed disabled person, by solving the problems of the lack of person’s social skills, adaptation at the workplace, and motivation for work, intermediation between the person and the employer as well as counselling by solving problems related to employment relations or conditions, and other assistance seeking more efficient and long-term integration in the labour market. The objective is continuous employment of the person for at least 6 months – in this case an institution providing vocational rehabilitation services will be paid for the service of support at the workplace. Moreover, seeking to thoroughly regulate the requirements for the content of the cycle of vocational rehabilitation services, provision of services, specialists who provide these services, and having regard to the nature of disability of persons participating in the Programme, on 6 January 2016, the Minister of Social Security and Labour approved Order No. A1-3, whereby the following were approved:

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- The methodology of provision of vocational rehabilitation services to persons who have visual, hearing, intellectual, mental and physical disabilities;

- General requirements for specialists who provide vocational rehabilitation services to the disabled. Persons participating in the Vocational Rehabilitation Programme are granted and paid the vocational rehabilitation benefit, irrespective of other income.

Statistics of the Vocational Rehabilitation Programme 2012–2015

	2012	2013	2014	2015	Plan for 2016
Funds used for vocational rehabilitation services	EUR 2 922.7 thous.	EUR 2 418.9 thous.	EUR 1 419.1 thous.	EUR 1 633.4 thous.	EUR 2 433.0 thous.
Funds used for vocational rehabilitation benefits	EUR 680.6 thous.	EUR 606.7 thous.	EUR 438.6 thous.	EUR 884.2 thous.	EUR 938 thous.
Number of persons who participated in the Programme	9020	896	626	711	750
Number of persons who completed the Programme	597	529	425	379	450
The employment rate within 6 months after completion of the Programme (percentage)	57	61	65.7	57.09	58

Data of the Lithuanian Labour Exchange under Table 3 the Ministry of Social Security and Labour.

On 1 March 2015, the Lithuanian Labour Exchange launched a new three-year project entitled "Assistance for the Disabled", this project was co-funded by the European Union Structural Funds and the state budget of the Republic of Lithuania. The project aims to increase the employment rate of the disabled through the provision of vocational rehabilitation services. The project will provide services for 2 000 disabled persons. The project value amounts to EUR 7 588 166.13.

Social Services

According the Law on Social Services (hereinafter referred to as the "Law"), social services are services aimed at providing assistance to a person (family) who, by reason of his age, disability, social problems, partially or completely lacks, has not acquired or has lost the abilities or possibilities to independently care for his private (family) life and to participate in society. The aim of social services is to create conditions for a person (family) to develop or to enhance the abilities and possibilities to independently solve his social problems, maintain social relations with society as well as to assist in the overcoming of social exclusion. It should be provided with a view to preventing the social problems of a person, family and community as well as to ensuring the social security of society.

The right to get social services has citizens of the Republic of Lithuania, aliens, including stateless persons, holding a permanent or temporary residence permit in the Republic of Lithuania and other persons in the cases provided for in international treaties of the Republic of Lithuania.

Provision of social services is based on personal needs of every person who requires getting help and is not based on disabilities, diseases or other factor as such. A complex assessment of a person's dependency is carried out according to the person's age, organism's functional disorders, social risk and the abilities related to these factors as well as motivation to solve one's social problems and the family's possibilities to care for the person, other peculiarities influencing the person's ability to care for his private (family) life or to participate in society. The need for social services is assessed the social workers appointed in accordance with the procedure laid down by a municipal institution.

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Social services are not sorted by clients' disabilities, diseases, age or other factors, but some organizations have specialization with what client groups they work. All social services are discussed with a client and individual plan is made according to that client personal needs.

Orphan Medicinal Products

32 orphan medicinal products by brand name were available in Lithuania during the period of 2015-2016 (August 15). 11 out 32 OMPs are introduced into the List of Reimbursed Medicinal Products or are centrally purchased by the State.

The orphan medical products used for the treatment of the ultra-rare diseases are reimbursed on the individual basis upon the request of the physician.

The special committee acting at the National Health Insurance Fund is responsible to take the decision on reimbursement of such orphan medical products without delay. The orphan medical products used for the treatment of the ultra-rare diseases are reimbursed on the individual basis upon the request of the physician. Accelerated pricing process is applied in that case.

Rare Disease Day

Every year Lithuania together with other countries mention RD day. The Ministry of Health each year draws attention of the citizens about RD issue, innovations providing medical care etc.

On the 10th of November 2014 in the Parliament Health care committee organised discussion entitled "What awaits a person in Lithuania's diseased pancreas neuroendocrine tumour?".

Also university hospitals organised events to mention RD:

1. Coordinating centre of Children's Rare Diseases in Vilnius University Hospital Santaros Klinikos organised annual international conferences for Rare Diseases day:

- 28th of February 2014 – 3rd scientific and practical conference "Rare Disease Day in Lithuania 2014: cystic fibrosis";
- 24th of October 2014 – School for children suffering from spina bifida and hydrocephalus and their families: how can we help?
- 20th of March 2015 – 4th international scientific practical conference "Lysomal storage diseases";
- 25th of February 2016 – International educational workshop on "Primary and Secondary Haemophagocytic Lymphohistiocytosis";
- 26 of February 2016 – 5h international scientific practical conference "The immune system and rare Diseases".

2. Coordinating centre for Rare Diseases of Hospital of Lithuanian University of Health Sciences Kauno klinikos activities for Rare Diseases day:

- Annual multidisciplinary open conferences are organized by the Coordinating centre for Rare Diseases of Hospital of Lithuanian University of Health Sciences Kauno klinikos dedicated to Rare Disease Days and targeted at multidisciplinary specialists and patients, with patient representative presentations (announced at EURORDIS website in 2016, the official EURORDIS Rare Disease video being translated into Lithuanian and sent to EURORDIS).
- Nation-wide and region-wide conferences for dermatologists, endocrinologists, ophthalmologists, neurologists, paediatricians, general practitioners, etc., either specifically dedicated to RD or RD included into the general agenda, organized by specialists of specific Rare Disease centres of the Hospital of Lithuanian University of Health Sciences Kauno klinikos in cooperation with specialist societies and patient organizations.
- Multiple mass media interviews are organized, with participation of specialists and patients. Presentations for awareness of RD for society sporadically or dedicated for RD Day (e.g., press conference at the Parliament of the Republic of Lithuania on the organizational aspects in the

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management of neuroendocrine tumours in Lithuania and on hypophysis diseases; different interviews publications in mass media.

Children‘s rare disease patients association event on the 4th of June 2016 “Let‘s not be alone with a rare disease“.

Other

Lithuania has accomplished many rare disease related achievements since the beginning of 2014:

- At the end of 2015 E-health system was launched which also contains information about rare diseases;
- In order to strengthen the availability and quality of the health care services Universal neonatal screening for congenital metabolic diseases procedure was updated (Order of the Minister of Health 2014-05-22, No V-601);
- In order to improve palliative care availability Palliative care services for adults and children procedure was updated (Order of the Minister of Health 2015-08-13, No V-946);
- In order to increase the availability of medicines, medical aid devices for people with rare disease Pharmaceutical Law was updated. Since 1st of January 2016 the definition of ultra-rare diseases was introduced.

The main areas of improvement since 2014 have been:

- The improvement of inter-sectoral cooperation;
- The strengthening of the early diagnosis of rare diseases through the uniting efforts of the health care institutions, health care professionals, NGO’s and participating in ERNs;
- The strengthening multidisciplinary approach for patients with rare diseases ;
- The Improvement of funding for rare disease diagnosis and treatment.

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