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RARE DISEASES IN THE SLOVAK REPUBLIC
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NATIONAL HEALTH INFORMATION CENTRE TASK REGARDING THE DATA AVAILABILITY ON RARE DISEASES IN THE SR

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Effective management of any group of diseases in the population, including rare diseases, needs to know the basic epidemiological parameters. The issue of rare diseases in the concept of state health policy has absented in many countries due to the difficulty of diagnosis, treatment and subsequent care of such patients. There is no conception of collection and processing of data regarding rare diseases, neither is there any unified conception of diagnosis, treatment, and overall health care of patients with rare diseases. The central availability of data concerning rare diseases is very low for different medical specialisations which deal with this issue. There exist only partial pieces of information, which are spread across different workplaces. The SR should participate in European information network on rare diseases. The SR is a member of ICBDSR (http://www.icbdsr.org) but not EUROCAT (http://www.eurocat-network.eu/), which is necessary to be changed!
The National Health Information Centre (NHIC) is a source of rare diseases in the following databases: National register of congenital anomalies, National cancer registry, Registry of patients with diabetes mellitus 1, National register of congenital anomaly of heart. The key to the knowledge of the real state of disease incidence rate in the population is an effective collaboration between relevant partners (including implementation of systematic control mechanisms), which is a basis for success.

Keywords: *National Health information centre – rare diseases – registries*

To effectively manage any group of diseases in the population, including rare diseases, it is inevitable to know the basic epidemiological parameters. The key moment enabling us to know not only the local, but also the wide-area incidence of the relevant group of diseases should be the interest of expert communities (often based on the activity of relevant authorities within them) and their central reporting.

Inclusion of the issue of rare diseases in the concept of state health policy has been absent in many countries due to the difficulty of diagnosis, treatment and subsequent care of such patients. Consequently, the rare diseases within the second Action Plan of Health Community 2007-2013 continued to appeal for the realisation of the relevant national program. The SR is finalizing the preparation of individual steps in solving this issue. The basis for a successful progress is to analyse the current state.

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The National Health Information Centre (NHIC) is a source of rare diseases in the following databases:

- National register of congenital anomalies-NHIC has at its disposal the data on congenital anomalies dating from 1965, which were processed within the federation separately for the Czech and Slovak Republic until 1992. After the end of the federation, the SR has been collecting this data separately.

Reports on congenital anomalies were for a long time focused on somatic mutations, less on metabolic disorders, which was related to the diagnostic availability and reporting duty of physicians. The rate of reporting, including rare diseases, was low since the general practitioners for children and adolescents who received the positively confirmed results sent by Screening Centre in Banská Bystrica, did not additionally report these results to NHIC, although the reporting duty concerned children up to 1 year of age until 2011.

The change of the system of reports to a register in terms of collecting data on congenital anomalies was a very positive change. Singular identification of these cases enabled solely by the register was a great contribution.

- National cancer registry- provides information on rare cancers and hemato-oncological diseases.
- Registry of patients with diabetes mellitus 1. Type - registers also newborns with diabetes.
- National register of congenital anomaly of heart- registers also rare congenital diseases of heart.

The most important source of data for the future should be the Registry of patients with congenital anomalies. However, it is necessary for individual clinical and laboratory workplaces to accept also changes in the system of congenital anomalies registration.

Since 2012, the reporting duty of congenital anomalies (according to the model of the Czech Republic) has been extended with reports of cases up to the age of 15. The reporting duty was thus assigned also to general practitioners for children and adolescents who should take the burden of reporting the congenital anomalies of specialists. The report form concerns mainly epidemiological data (including prenatal diagnostics). It is possible that in the future, congenital anomalies reporting without reference to the age will be adopted.

In order to control the data on rare diseases (in terms of available databases), an effective collaboration of NHIC with a Newborn Screening Centre in Banská Bystrica is necessary (in addition to the regular communication with clinical and outpatient physicians). In addition, it is beneficial to cooperate also with health insurance companies, which register also cases which require specific treatment for individual diagnoses. The clinical registries, which are important for physicians from the point of view of clinical state monitoring, treatment, and quality of patients' lives, should be in full competence of the relevant expert centres.

The key to the knowledge of the real state of disease incidence rate in the population is an effective collaboration between relevant partners (including implementation of systematic control mechanisms), which is a basis for success. We hope that the inter-branch working group for congenital anomalies in the SR reaches its goal.

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ÚLOHA NÁRODNÉHO CENTRA ZDRAVOTNÍCKYCH INFORMÁCIÍ V DOSTUPNOSTI ÚDAJOV O ZRIEDKAVÝCH CHOROBÁCH V SR

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Pre efektívne manažovanie akejkoľvek skupiny chorôb v populácii, vrátane zriedkavých chorôb, je nevyhnutné poznanie základných epidemiologických

parametrov. Zaradenie problematiky zriedkavých chorôb v koncepcii štátnej zdravotnej politiky vo väčšine krajín, pre náročnosť diagnostiky, liečby i následnej starostlivosti o takýchto pacientov, absentovalo. Tak ako neexistuje jednotná koncepcia diagnostiky, liečby a celkovej starostlivosti o pacientov so zriedkavými chorobami, neexistuje ani koncepcia zberu a spracovania údajov o týchto ochoreniach. Pre rôzne medicínske špecializácie, ktorých sa predmetná problematika dotýka, je centrálna dostupnosť údajov o zriedkavých ochoreniach nízka. Existujú iba čiastkové informácie, ktoré sú roztrieštené po rôznych pracoviskách. SR by mala byť zapojená aj do európskej informačnej siete o zriedkavých chorobách. The SR should participate in European information network on rare diseases. NCZI je členom v ICBDSR (medzinárodný ústav vrodených chýb - http://www.icbdsr.org), ale nie v EUROCATe (európsky dohľad vrodených chýb - http://www.eurocat-network.eu/), čo je potrebné zmeniť!
Národné centrum zdravotníckych informácií (NCZI) je zdrojom informácií o zriedkavých chorobách v nasledujúcich registroch: *Národný onkologický register – Register pacientov s diabetes mellitus 1.typu, Národný register vrodených chýb srdca* – eviduje aj zriedkavé vrodené chyby srdca. Kľúčom k poznaniu reálneho výskytu chorôb v populácii je efektivita spolupráce medzi príslušnými partnermi (vrátane zavedenia systémových kontrolných mechanizmov).

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