

Rare Diseases Management Optimization in Children and Adolescents in Georgia
(Plan of Actions)

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Abstract. The practitioners in Georgia are not well-acquainted with the rare diseases, and it predetermines the prescription of a multitude of unnecessary and potentially hazardous invasive diagnostic interventions. Because of the insufficient implementation of the evidence based medicine principles as well as the absence of special algorithms, the correct diagnosis is belated; quite frequently it is not made at all. Therefore the management of these diseases can not be considered as adequate and efficient and this fact is jeopardizing in particular the children and adolescent health. The establishment of informational referral centers for rare diseases is considered to be an effective response to this challenge.

The main goals for optimizing the management of the rare diseases in children and adolescents were identified as follows: selection of the rare diseases being current in Georgian children and adolescent population employing the Delphi and Nominal groups technique; creation of the register of patients (children and adolescents) with rare diseases; creation of the management algorithms (expert systems) for the clusters of the rare diseases employing the up-to-date information technologies and new approaches (fuzzy logic); organization of the pediatric informational referral center for the rare diseases; collaboration with similar centers worldwide. The obtained materials could be used as a database for health care management, for conducting a pre-marketing research as well as clinical trials in order to define the efficiency of corresponding medicines.

Importance of the Problem, Research Novelty

The nosologic units with prevalence not exceeding 0.05% of general population belong to the group of the rare diseases. There are several thousand rare diseases. Their total number is quite significant as 5-10% of population have some rare disease [1, 2, and 3]. The specific weight of the rare diseases in children and adolescent morbidity structure is increasing all over the world, particularly in the developed countries. As to the countries with transitional economics, including Georgia, the information of this kind is hardly available.

The practitioners, at least in Georgia, are not well-acquainted with the rare diseases, and it predetermines the prescription of a multitude of unnecessary and potentially hazardous invasive diagnostic interventions. Because of the rarity of this group of diseases and insufficient implementation of the evidence based medicine principles as well as the absence of special algorithms, the correct diagnose is usually belated; quite frequently it is not established at all. It is noteworthy, that according to the materials found beforehand, there is actually no pediatric software in the world, which could be applied to a purposeful diagnostics of clusters of various rare diseases. Equally, fuzzy logic approaches are not actually employed.

The treatment and, generally, the management of these diseases can not be considered as adequate and efficient, this fact particularly jeopardizing the children and adolescent health. It is also noteworthy that due to the rareness of the rare diseases, it is not financially reasonable for pharmaceutical companies to produce medicines to treat them; therefore, the relevant research and production are quite limited. Because of the limited number of cases it is quite difficult to establish the efficiency of the therapeutic interventions by employing the classical methods. This fact in its turn indicates the advisability of

elaborating new methods and means of treatment and assessing their efficacy as well the importance of the multi-center cooperation.

The establishment of informational referral centers for rare diseases is considered to be an adequate response to this challenge. The total world-wide number of these centers is quite low. Therefore, there exists only limited experience regarding their organization and functioning peculiarities.

The premises indicate the gravity of the problem and the necessity to find the ways to solve it not only in Georgia, but globally as well. The innovative nature of the presented research is defined by the following factors: a) Creation of diagnostic software for clusters of rare diseases in children and adolescents employing the up-to-date information technologies and new approaches (fuzzy logic); b) Establishment of the register of the rare diseases in children and adolescents in Georgia; c) Clarification of the foundation and functioning specifics of the pediatric informational referral centers for rare diseases in countries with transitional economy.

Goals and Objectives of Research.

The main goal of research is to optimize the management of rare diseases in children and adolescents.

Objectives (and tasks) are as follows:

- a) To select the rare diseases being current in Georgian children and adolescents
 - Create a list of rare diseases in children and adolescents
 - Establish a list of rare diseases being current in Georgia
 - Establish a list of top 50-100 rare diseases being most important in Georgia
- b) To create a register of patients (children and adolescents) with top 50-100 rare diseases
- c) To create diagnostic algorithms (expert systems) according to clusters of the diseases
- d) To organize the pediatric informational referral center for rare diseases
 - Look for the information regarding the informational referral centers for rare diseases
 - Create the necessary preconditions for the foundation of the center (office, political, administrative and community support)
 - Inform doctors, adolescents and their parents
 - Create the web-site
 - Initiate the international cooperation
 - Define the efficacy of the center work.

Compliance of Research methods with the Project Goals and Objectives

- Creation of the comprehensive list of the rare diseases with childhood or adolescence onset, based on the analysis of the information available from manuals, monographs, scientific periodicals, Internet and specialized databases (OVID, MEDLINE) and the meta-analyses of the data presented in the literature;
- Selection of the 50-100 top rare diseases being current in the country employing the Delphi and Nominal groups technique [4];
- Work on and analysis of the above-mentioned data in order to assess the information value of the diagnostic criteria of the clusters of the rare diseases;
- Creation and validation of management algorithms for the clusters of the selected rare diseases based on the obtained results and employing the principles of the evidence based medicine and fuzzy logic [5,6];
- Detection of children and adolescents with selected rare diseases and creation of the register will be performed based on the results of the comprehensive clinical and laboratory investigation of the patients taking into consideration the specifics of the epidemiological investigation;
- Organization of the pediatric informational referral center for rare diseases and establishing its efficiency employing the epidemiologic research methods.

Expected results of Research and their importance for science, economy and/or social sphere

The expected results of the research are the following: creation of the register of the rare diseases in Georgian children and adolescents, creation of the management algorithms for the clusters of the rare diseases which are the most important ones for the country, organization of the pediatric informational referral center for rare diseases and defining the specifics of its creation and functioning in a country with transitional economics. It is noteworthy that the expected results (except for the register of the rare diseases) are the priority ones.

The obtained results would facilitate the optimization of the rare diseases management in children and adolescents, particularly in terms of a substantial improve in the diagnostics and treatment. The project implementation would also provide a significant input in the area of employing the up-to-date information technologies and new approaches (fuzzy logic) in clinical medicine and particularly in pediatrics. In our opinion, it would be quite useful for further perfection of the children and adolescent healthcare system improvement.

The following categories of population should be considered the beneficiaries of this research: up to 10% of children population, who should have some rare disease, their family members and care-givers. From scientific point of view, the creation of the management algorithms based on new approaches as well as the receipt of new evidence in the area of the public healthcare, in particular, for resolving the rare diseases related problems, would be a great novelty. The obtained materials could be used as a database for healthcare management, for conducting a pre-marketing research and clinical trials by pharmaceutical companies in order to establish the efficiency of some medicines.

Acknowledgments. The research is founded by GNSF/ST08/6-460.

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