OPPORTUNITIES FOR RESEARCH AND MONITORING OF DISEASES WITH A GENETIC NATURE IN GENERAL PRACTICE

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ABSTRACT

As a "gatekeeper" in the health system, the family doctor is the first competent medical person, who could determine the existence of hereditary relation in the diseases of their patients. In this regard, we set the goal to determine personal position of general practitioners from Stara Zagora region on opportunities for research and monitoring of diseases with genetic character in outpatient primary outpatient care. In a pilot study, we found that a large percentage of family physicians are not informed about new, modern methods to detect and prove the hereditary nature of the disease in their patients. Additionally, family physician are prevented of using the potential of medical and diagnostic capabilities in research and monitoring of patients, in which etio- pathogenesis of the disease it is suspected for genetic relation.

Key words: GPs, diseases with a genetic nature, research, monitoring

INTRODUCTION

Realization of the genome project, the progress in medical genetics and biotechnology in the last decade (the discovery of the 32,000 genes in humans, proving of relationship between certain genes and disease processes, the development of pharmacogenetics and pharmacogenomics, the discovery of new therapeutic targets and new generation drugs and etc.) shifted the center of hereditary pathology of pediatrics and neonatology to all other specialties. (1) In this regard, GPs as specialists in general / family medicine are responsible not only and primarily for the early detection of pathological process and clinical diagnosis, but for the opportunely referral of patients to genetic laboratories to conduct adequate treatment and prevention of established hereditary diseases or predispositions. As a main, primary filter and guide to each patient in curent national health system, family medic daily faces with a number of patients in etio- pathogenesis which

disease has established a hereditary relation. In order to effectively fulfill its role and assumed responsibilities, GPs need advanced information on genetic diseases, as well as additional knowledge to establish proper and adequate behavior in consultation and subsequent medical and diagnostic activities for patients with suspected genetic components of newly discovered or already existing disease.

Goal:

The aim of our team was to analyze the opinion of a group of family doctors from Stara Zagora region, Bulgaria about difficulties, which GPs faces in consultation, diagnosis and monitoring of patients with hereditary diseases.

MATERIALS AND METHODS

To realize the goal, it was conducted a pilot study among 43 GPs from Stara Zagora region, Bulgaria, in the period from January to May 2014. According to National Statistics Institute of 31.12. 2013, on the territory of Stara Zagora region, covering the towns of Stara Zagora, Chirpan, Maglij, Kazanlak, Radnevo, Gourkovo and adjacent villages to municipalities, there practise 234 GPs. . (2) In the sample provided by us, the most numerous

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was the age group of doctors between 41 and 50 years- 53.5%. 20% of respondents have opened ambulatory for primary health care in villages and 77% of them - work mainly in cities. Family physicians completed a standard questionnaire including 44 questions with answers gated. In a questionnaire included questions concerning the level of awareness and knowledge of GPs on genetic diseases as well as opportunities for their early diagnosis by implementation a wider specialized and highly selective methods and tests in the general practice. In the survey are not mentioned personal data (name, date of birth), i.e. according to observing the principles of voluntariness, anonymity and correctness.

DISCUSSION AND RESULTS

Basic scientific research has led to a greater understanding of the contribution made by genes to present and future human health. (3, 6) It is increasingly recognized that genetic information will need to be integrated into all aspects of health care delivery, including primary care. (4, 5, 6) According to RCGP (Royal college of General Practitioners), it has been estimated that at least one in ten of the patients seen in primary care has a disorder with a genetic component. (7) Many foreign authors and researchers have ascertained that primary care providers have varying levels of involvement and confidence in genetics, just like variability in the quality of health care provided to patients with hereditary diseases. (8, 9, 10, 11, 12) Emphasis is placed on the need for GPs to have the appropriate level of knowledge in genetics and new technologies. (13, 14, 15, 16, 17) In this regard, in most European countries are involved a multiplicity of structures and organizations in training health professionals in genetics. (18, 19, 20, 21, 22, 25) For example according to their approved curriculum of ROYAL COLLEGE OF GENERAL PRACTITIONERS, there are three main themes of genetics in primary care: identifying patients with, or at risk of, a genetic

condition; clinical management of genetic conditions; communicating genetic information.

As opposed to other European countries, in educational programs for acquisition of specialty "General Medicine" in Bulgarian medical universities lack training module on Medical Genetics. Until now are not organized and courses for continuing medical education to improve the knowledge of providers of primary health care in genetics and genomics. Respondents from our team expressed the opinion, that they are less familiar and do not have updated information about new methods, testing and approaches in these medical fields. This is one of the reasons, family physicians to apply conventional clinical laboratory tests in clarifying the diagnosis of genetic diseases to their patients. Another important factor that determines the potential and range of health care in connection with the early diagnosis of patients with hereditary disease in general practices are introduced regulatory standards by National Health Insurance Fund. So due to limited opportunities in realization of forms for medical diagnostic activities and imposed limit by the contractual partner in the face of National Health Insurance Fund, GPs is forced to use more widely available specialized clinical diagnostic methods for patients suspected of diseases with hereditary etiology. Mostly as shown in **Figure 1**, these are clinical - laboratory, biochemical, X- ray and ultrasound tests. (Figure1) The percentage of respondents used genealogical analysis, DNA analysis and cytogenetic tests varies between 9.3% and 11.6 %. Most of the surveyed family doctors apply mainly biochemical blood tests which are less selective. And nearly half of respondents (46.5%) said that they hadn't used research to clarify the genetic nature of the disease or predisposition to a genetic disease to their patients. Unfortunately, the situation is similar in general practice in other European countries.

Figure 1.

In clarifying the diagnosis of genetic diseases, you are appointed following tests:	Percent:
Biochemical tests	32.56 %
Cytogenetic tests	9.30 %
Genealogical analysis	11.63 %
DNA analysis	11.63 %
X- ray and ultrasound tests	27.91 %
Others tests	23.26 %
No used research	46.51 %
No answers	4.65 %

According to Challen K. and colleagues their work has shown clearly that, apart from family history taking, many European GPs do not consider that "genetic" care should form part of their practice. (6) In the study conducted by us, family physicians were also adamant that suspected disease with a genetic component, appointment of highly selective research, interpretation of obtained results in connection with early diagnosis and successive therapy should be implemented by the relevant

narrow specialist. To the question "What kind of specialists you most often refer your patients for diagnosis in suspected disease that is genetically determined?", most of the respondents have indicated more than one specialist depending on the type of disease affected functional system (endocrinology, obstetrics and gynecology, neurology and others.) and according to the age of the patient (preferably Pediatrics- in newborn, neonates, infants and children). (**Figure 2**)

Figure 2.

What kind of specialist most often you refer your patients for diagnosis?	Percent:
Pediatrics	55.81 %
Obstetrics and gynecology	25.58 %
Endocrinology	60.47 %
Neurology	18.60 %
Others	20.93 %
No answers	6.98 %

All family physicians participating in the study had an opinion that mandatory patients which diseases are hereditary conditioned, they should be counseled and monitored their condition in terms of therapeutic benefit / impact in particular specific treatment from relevant specialist in higher levels of health care and with more depth knowledge and experience. (**Figure3**) According to the frequency of nosologic units, which are encountered in their general practices, family doctors think that patients with genetic pathology need more and with larger range of

qualified health care (medical examinations, consultations, prevention, therapy, etc.) provided by pediatricians, otorhinolaryngologists, ophthalmologists, neurologists, endocrinologists, etc. In addition, all people who live with a genetic condition (either personally or in their family) should have access to appropriate, up-to-date and accurate information. Additionally, they should have available the necessary support to assist them to manage the challenges to their health and wellbeing and to enable them to reach their full potential. (22)

Figure 3.

What medical specialists your patients with hereditary diseases have the greatest need in terms of medical examinations and monitoring?	Percent:
Pediatricians	46.51 %
Otorhinolaryngologists	4.65 %
Ophthalmologists	6.98 %
Neurologists	25.58 %
Endocrinologists	51.16 %
Gynecologist	11.63 %
Others	23.26 %
No answers	11.63. %

On the one hand, from the family doctor is expected to be competent and has a wide range of knowledge and skills in all medical disciplines, respectively medical genetics. At the same time, at the national level lack approved training module in curricula for specialty "General Medicine". Family doctor did not receive updated information about newly introduced genetic technologies,

methods and tests, as well as opened and active genetic centers in the country where to refer their patients, who need of highly qualified medical care. A relatively small group of family physicians included in the pilot study are aware of and had set specialized genetic tests as DNA analysis, genealogical analysis and others. On the other hand the potential of opportunities for GPs to quickly and early

diagnosis of genetic disease or predisposition by highly specialized and expensive technologies and research is limited by the National Health Insurance Fund. The appointment of this type of specific research leads to financial sanctions for the family doctor by the contractual partner (NHIF). The package of medical services performed by providers of primary outpatient medical care, according to the National Framework Agreement and regulatory requirements of the NHIF lack of appointment of genetic testing and interpretation of received results. In legal documents reviewed by us, it is clear that in conjunction with implementation of prevention and promotion of health at the level of primary health care, from family doctor is required to wary to the presence of risk factors, including the so- called "unmodified" to which belong and genetic factors, and also to explore significant family history to assess potential genetic risk of altered health states. (23, 24)

In addition, the family doctor is obliged conduct individually or in teams with other professionals and specialists premarital medical consultation and family planning, to identify and evaluate risk factors with individualized adjustments, to inform to refer and to include in national programs for health promotion, to prepare individual and family educational programs on the effects of risk factors and possibilities for their correction. (23, 24) Analyzed regulatory requirements determine the need for a comparatively extensive and thorough training of the family doctor in the medical discipline "Genetics". Such a wide range of knowledge and skills need to possess and practise every European family doctor. That is why a number of training programs in genetics for GPs focus on the acquisition and development of key competencies in this area, which have a great practical value in working with patients with suspected or already proven genetic disease in practice for primary care. For the authors Challen K. and team, it is clear that given the significant effect of country of practice, independent of all other factors, on practitioner behavior, recommendations on genetic education at all levels will have to be sensitive to country-specific issues. And also educational structures and content will require tailoring to local priorities and learning conventions. (6)

CONCLUSION

As the first point of contact for patients, GPs fill a certain 'gate-keeping' role. In this sense, their direction regarding specialist and support interventions is very important. (22)

Based on our conducted pilot study, we make the following conclusions:

- A large percentage of family physicians are not informed about new, modern methods of detection and prove the hereditary nature of the disease in their patients.

- Additionally, general physicians are prevented for using the potential of medical and diagnostic capabilities in research and monitoring of patients in which etio-pathogenesis of the disease is suspected a relation.

Rapid advances in genetics have led to major impacts upon general practitioners' (GPs) needs to better understand and provide information and referrals on genetic conditions to patients and their families. (22) Genetics in general practice involves managing diseases that run in families. The management of genetic diseases and conditions involves both the individual and their family. This determines, in the process of clarification and early diagnosis of patients with diseases that have a genetic predisposition, GPs to be aware and fully informed of hereditary pathology of the disease, their way of detection and identification.

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