

SCIENTIFIC STATEMENT

By assoc. prof. Trifon Georgiev Chervenkov, MD, PhD – associate professor at Department of Medical Genetics, Medical University “Prof. Dr. Paraskev Stoyanov” – Varna.

Regarding: dissertation thesis for obtaining academic and scientific degree “Doctor of Philosophy”, in scientific speciality “Genetics”, professional field 4.3. “Biological Sciences”, Higher education area 4. “Natural sciences, Mathematics and Informatics” of Milena Petrova Stoyanova, MD, doctoral student at Department of Medical Genetics, Medical University “Prof. Dr. Paraskev Stoyanov” Varna, on the topic “*Genetic Diagnostic Study among Pediatric Patients with Hereditary Pathology Who Received Genetic Counselling*”, with research supervisor Prof. Lyudmila Boncheva Angelova, MD, PhD.

I have been appointed as a member of the scientific jury of the dissertation thesis of Dr. Milena Petrova Stoyanova with Order of the Rector of Medical University Varna № P-109-329/01.08.2022, based on resolution of the Faculty council of the Medical Faculty of the Medical University Varna.

I. Common remarks for the dissertation thesis of Dr. Milena Stoyanova.

The dissertation thesis of Dr. Milena Stoyanova is correctly structured and encompass 149 standard print pages. The text is distributed in 12 chapters. The literature review, the acquired data and the analyses done are visualized with 17 tables and 34 figures. Bibliography contains 246 sources, 25 in Bulgarian language and 221 in English language.

II. Assessment of the actuality of the topic

The dissertation thesis’s topic is with ever increasing actuality at the present moment:

subject is medico-genetic consultation of hereditary diseases and congenital anomalies, which are rare diseases as separate entities, but in sum they affect in-between 4% and 7% of the population and represent a burden not only for the patient, but also for its family and the society as a whole. Genetic consulting is a major approach to prophylaxis of hereditary diseases, but in the last years its actuality constantly increases with the increasing abilities to establish the exact genetic diagnosis, and also due to the increasing implementation of new methods of treatment, including definitive gene therapy. In addition, the existence of scarce information for the functioning of genetic consultation in our country, the topic “*Genetic Diagnostic Study among Pediatric Patients with Hereditary Pathology Who Received Genetic Counselling*” is exceptionally actual.

III. Assessment of the literature review

The literature review is comprehensive – it introduces the reader to the issues of the dissertation, but at the same time it is also balanced: it occupies about a quarter of the dissertation work. In practice, the literature review includes four parts that describe hereditary and congenital pathology in childhood, the methods used in the diagnosis of children with suspected hereditary pathology, a description of the highly specialized activity of medical genetic counseling and a summary of the presented literature data, and it naturally leads to the set aim and objectives.

IV. Assessment of the set aim and objectives

The main aim of the dissertation work is clearly formulated: descriptive-epidemiological assessment and study of the effect of genetic counseling as an approach to clinical-genetic diagnosis in persons with suspected hereditary pathology in childhood based on the experience of the University Genetics Center over a period of ten years. To achieve the goal, five objectives have been formulated, which are well-founded to achieve the aim. Given the methods presented, this aim is feasible.

V. Assessment of the materials and methods used

The presented clinical cohort is impressive and includes 3,124 childhood patients referred to the Laboratory of Medical Genetics of University Hospital Saint Marina Varna with various indications. From them, a group of 968 patients was selected, to whom an active diagnostic approach (laboratory and consultative) was applied to make the diagnosis (clinical and/or genetic). The clinical and laboratory methods used, the used dysmorphological databases and software online-based programs and methods for statistical processing of the data are described in detail. The results obtained are competently discussed.

VI. Assessment of the presented results and conclusions

The results are illustrated with convincingly presented tables and graphs. They can be summarized in two directions: descriptive - epidemiological characteristics of patients, and results of laboratory and clinical research - conventional cytogenetic analysis, DNA analysis for selective screening, diagnostic-consultative activity. The results are competently discussed in the light of the literature review on the problem and show an excellent knowledge of both the clinical and laboratory aspects of the topic by the PhD student. Five conclusions were drawn, which summarize the obtained results and are in accordance with the set aim and objectives of the doctoral studies.

VII. Assessment of the contributions of the doctoral thesis

I accept the contributions of the doctoral thesis, as the work fills a gap in our country through a systematized descriptive-epidemiological characterization and evaluation of the role of the active medico-genetic activity of a structure in Bulgaria. In addition, on this basis, the most effective indications for guiding the search for the genetic diagnosis can be derived.

I define the contributions as fully sufficient for the acquisition of the academic and scientific degree “Doctor of Philosophy”.

Conclusion

The dissertation thesis of Dr. Milena Petrova Stoyanova examines a current topic about the practical aspects of medical genetic counseling in our country. In my opinion, this is a profound scientific development, meeting the requirements of the law for academic growth of the Republic of Bulgaria and the academic growth regulations of the Medical University Varna for the acquisition of the academic and scientific degree “Doctor of Philosophy”. That is why I will vote positively and allow myself to recommend the same to the other members of the respected Scientific Jury of the procedure.

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Varna


/assoc. prof. Trifon Chervenkov, MD, PhD/