МЕДИЦИНСКИ УНИВЕРСИТЕТ - ВАРНА
„Проф. д-р Параскев Стоянов“
Ул.“Марин Дринов“ 55, Варна 9002, България
Тел. : 052/ 65 00 57, Факс: 052/ 65 00 19
e-mail: uni@mu-varna.bg, www.mu-varna.bg

MEDICAL UNIVERSITY - VARNA
“Prof. Dr. Paraskev Stoyanov”
55, Marin Drinov Str., 9002 Varna, Bulgaria
Tel.: +359 52/ 65 00 57, Fax: + 359 52/ 65 00 19
e-mail: uni@mu-varna.bg, www.mu-varna.bg

Fund “Nauka” Project № 19035 Resume – Competition-Based Session 2019:
“Study of balanced subtelomeric rearrangements by molecular cytogenetic analysis in patients with combined reproductive problems”
Project leader: Prof. Lyudmila Boncheva Angelova, MD, PhD

The purpose of the study is to demonstrate and evaluate the role of subtelomeric rearrangements in patients who visited the Laboratory of Medical genetics, University Hospital “St. Marina”, Varna, due to reproductive failure – miscarriage (1 or more) in combination with another reproductive problem (stillbirth, affected children with congenital anomalies with/ or without mental retardation). The identification of balanced “hidden” subtelomeric translocations would be helpful in establishing the etiological causes of couple‘s reproductive problems by discovering the genetic mechanism responsible for them. In this way, families will be provided with more precise genetic counseling (evaluation of recurrent risks of subsequent unsuccessful pregnancies, prenatal diagnosis and prevention of affected offspring). This illustrates the importance of searching such rearrangements in couples with reproductive problems of uncertain etiology.

The molecular cytogenetic method – subtelomeric FISH, will be used to screen the subtelomeric regions, to search for balanced “hidden” subtelomeric rearrangements in patients with a normal karyotype by conventional cytogenetics. This is an innovative method for our country, which will allow us to evaluate and analyze the factors for reproductive failure in the Bulgarian population in more details and compare them with data from similar approach.

The study will not only improve genetic counseling process for infertile couples but will implement a new type of molecular-genetic method in the laboratory practice of our division.

The study will be useful for future research and will give a new perspective of reproductive failure in general.

This illustrates both the scientific and practical contributions of this type of research.

Expected results:

The type, presence and frequency of conditionally balanced, subtelomeric (hidden) chromosomal aberrations in families with combined reproductive problems in our country were determined, as well as the probability of giving birth to healthy offspring in a couple with an established chromosomal aberration.

The results of the study are an attempt to improve medical and genetic counseling in couples with reproductive failure and the introduction of a new type of molecular genetic method in laboratory practice – FISH – to search for "hidden" changes, as one of the possible reasons for failure reproduction in a pair.

The results of the research are summarized in the dissertation of the doctoral student Maria Kiryakova Tsvetkova.

Results:

Of the patients included in the study, analysis with subtelomeric FISH could be performed in 20 patients from a total of 85 selected individuals meeting indications for a possible diagnostic result from molecular cytogenetic analysis – patients with combined reproductive disorders who showed normal karyotype in conventional cytogenetic method. These are patients with preserved cell suspension for the last 3 years, or others who may have been contacted and responded to the additional study (after signing an informed consent). In this group 1 subtelomeric chromosomal disorder was found – 5% (1/20).

This is a couple with a child born with chromosomal disease – 46, XX, der (4). This result recommends additional analysis in parents by subtelomeric cytogenetics. Subtelomeric rearrangement was found in the father between chromosomes 4 and 6 and a normal result in the mother.