Vol 5 | Issue 4 | 2015 | 218-221.

e-ISSN 2248 – 9142
print-ISSN 2248 – 9134

International Journal of
Current Pharmaceutical & Clinical Research

www.ijcpcr.com

FREQUENCY OF CHROMOSOMAL TRISOMIES AND RISK FACTORS

Danielius Serapinas*

Mykolas Romeris University, Lithuania.

ABSTRACT

The aim of the study is to evaluate the use of cytogenetic and molecular cytogenetic techniques to prenatal trisomies setting 13 , 18 and 21 of chromosomes for patients during the 2007 to 2011, that had genetic counseling to determine between these trisomies and risk factors.1,156 patients were examined and for 49 (4.2%), abnormal karyotype was identified, of which 30 (61%) are associated with Down syndrome, 14 (29%) to the Edwards syndrome and 5 (10%) are those relating to Patau syndrome. After studying questionnaires and case records data, based on the statistical reliability of the data analysis of fetal abnormality association with the main factors on reproductive (abortions, contraceptives) and other risks or environmental factors (smoking, genetic diseases and drug abuse) was performed.

Key words: Chromosomes, Genetic testing, Prenatal.

INTRODUCTION

Congenital disorders very often leads to physical development defects and intellectual disability. Therefore, all neonates who have certain congenital abnormalities (including external and internal organs), or which are intellectually disabled chromosomes should be analyzed. Congenital disorders can be diagnosed in any organism development (prenatally and after birth) period. If there are members familywith such diseases is recommended for prenatal diagnosis, allowing diagnosis of the disease before the child's birth. [1]. Most oftenly, a variety of chromosomal changes in fetal tissues noticed premature chromosome. 15-20% of all pregnancies to be terminated, that is, abortion and pregnancy outcome of which about half of these miscarriages are associated with chromosome number or structure errors, which is incompatible with life. Various chromosome abnormality is diagnosed in approximately for 6-7 newborns in 1000, which is approximately 0.7% of newborns. The most common chromosomal abnormality is a chromosomal changes associated with a number aneuploidies are 21 th, 18 th and 13 th chromosome. This work presents a congenital disorders that are diagnosed with the genetic laboratory analysis, clinical chemistry and laboratory genetics. Syndromes are such: 1. Downsyndrome (trisomy of chromosome 21); 2. Edwards syndrome (trisomy of chromosome 18); 3. Patau syndrome (trisomy of chromosome 13).

The diagnosis is based on non-invasive methods of blood serum biochemical indicators during the investigation of the first and second trimester of pregnancy, and medical ultrasound. Biochemical analysis of blood serum is an important non-invasive method - PRISCA that helps defect patients who face a higher risk of congenital anomalies. Modern serum analysis helps to identify pregnant women who face a higher risk neutral tube defects and trisomies of chromosomes 21 and 18 (Down syndrome and Edwards syndrome) [2].

Depending on the age of the woman, her medicine, obstetrics, family history and if higher risk of chromosomal anomalies observed Amniocentesis is recommended to perform an invasive test that helps diagnose chromosome number and structure anomalies and examining -The whole chromosome karyotype and performing certain FISH tests [3].

Corresponding Author :- **Danielius Serapinas** Email:- dserapinas@gmail.com

According to studies, the only obvious feature is the forementoned aneuploidies and maternal age connection: the risk to give birth to a child with Down's, Edwards and Patau syndromes is much higher for women who are 35-40 years old, compared with younger women [4-6].

MATERIALS AND METHODS

The study consists of the following stages (Figure 1):

Stage 1. Prisca non-invasive tests are carried out. During the first and second trimesters of pregnancy serum biochemical indicators and ultrasound medical analysis is performed. These indicators help identify pregnant women who are more likely to give birth to a newborn with different chromosome abnormalities .

Stage 2. Patients who have a higher risk Prisca test results, consult a geneticist.

Step 3: Patients are offered opportunities for further study, with a diagnosis of prenatal diagnostic method - Amniocentesis.

Step 4. FISH tests are carried out. Some positive results are obtained, karyotype is a proven method using a karyotype test. The analysis phase is analyzed in this study.

The study material of the fourth phase of analysis of fetal cells in the amniotic fluid, which provide information about fetal chromosomes. Amniotic fluid is taken from the uterus during 16-22 weeks using invasive prenatal diagnosis (amniocentesis out method). Medical procedures determined by the physician-obstetrician. A thin needle into the pregnancy the amniotic sac of fluid within the stomach and womb lining. After the amniotic fluid (about 10-15 ml) into two disposable syringes, needles are taken out. Amniotic fluid should be free of blood supplements. Within one hour of collection sample delivered to the laboratory. The temperature should be 18 - 25 ° C .

Amniocytic that were collected in the laboratory is divided into two parts: amniocytic for molecular cytogenetic FISH test (5-8 ml), which should help to identify specific chromosome with its specific symptom and 5-10 ml amniocytic this activity cytogenetic tests, which should help establish full karyotype.

RESULTS

In order to diagnose chromosomal abnormalities prenatally by using molecular cytogenetic method FISH and cytogenetic karyotype test method (analysis system Cyto Vision Leica Biosystems, Germany) study material amniocytic 1,156 patients were tested. In research studies, which were conducted during 2007 - 2011 years. This figure includes 4.2% of patients who were admitted to a risk group, that is, every 24 female fetus was diagnosed with a chromosome abnormality, which is linked down (47, XX, +21; 47, XY, +21), Edwards (47 XX , 18; 47, XY, 18) and Patau (47 XX, 13; 47, XY, 13) syndromes. 30 patients of the study group (1156 patients) was diagnosed with Down syndrome (2.6%), 14 were diagnosed with Edwards Syndrome (1.3%) and 5 with (Patau syndrome (0.4%). (Table 1).

Of the 32 patients, who during this period gave birth to newborns with aneuploidies of +13, +18, +21 chromosome, card cases and questionnaire analysis was performed. In addition, according to case records and questionnaires of 40 patients, who belonged to the control group Analysis was carried out. In the above-mentioned patients were considered invasive tests.

Having analyzed the data and analysis of cases of card data and then processing these data with statistical package of social sciences. Referring to the statistical reliability ($p < 0.05$ is completely reliable), the basic reproductive factors (the abortion pill) or environmental factors (smoking, genetic diseases and drug use) influence fetal anomalies were found.

Risk and environmental factors that affect fetal anomalies such as smoking were analyzed. The results are presented in Figure 2. After analyzing the questionnaires, it was observed that 3% of patients in the pathological groups used tabacco products after fertilization, 13% used to smoke before pregnancy, and 84% of patients who did not use tabacco products. Meanwhile, patients in the control group have not been used tabacco at all, and 3% of patients used to smoke just before getting pregnant. Composes 97% of patients who had never smoked. According to statistics releability that tabacco use is very informative risk factor that is associated with a diagnosis of anomalies.

Table 1. **The frequency of diagnosed syndromes in 2007 – 2011**

Year	2007	2008	2009	2010	2011	In total
Syndrome	**n / %**	**n / %**	**n / %**	**n / %**	**n / %**	**2007-2011m.**
Down syndrome	4 / 2.9%	3 / 1.3%	9 / 2.6%	5 / 2.3%	9 / 4.1%	30 / 61%
Edwards syndrome	2 / 1.5%	5 / 2.1%	3 / 0.87%	3 / 1.4%	1 / 0.45%	14 / 29%
Patau syndrome	0 / 0%	1 / 0.4%	2 / 0.58%	0 / 0%	2 / 0.9%	5 / 10%
The total number of investigatives	136	237	345	216	220	1154
The total number of pathologies	6 / 4.4%	9 / 3.8%	14 / 4.1%	8 / 3.7%	12 / 5.5%	49 / 4.2%

n – diagnosed trisomy;

% –proportion of mutations , many new cases of disease are diagnosed during a year

Figure 1. The scheme of study design.

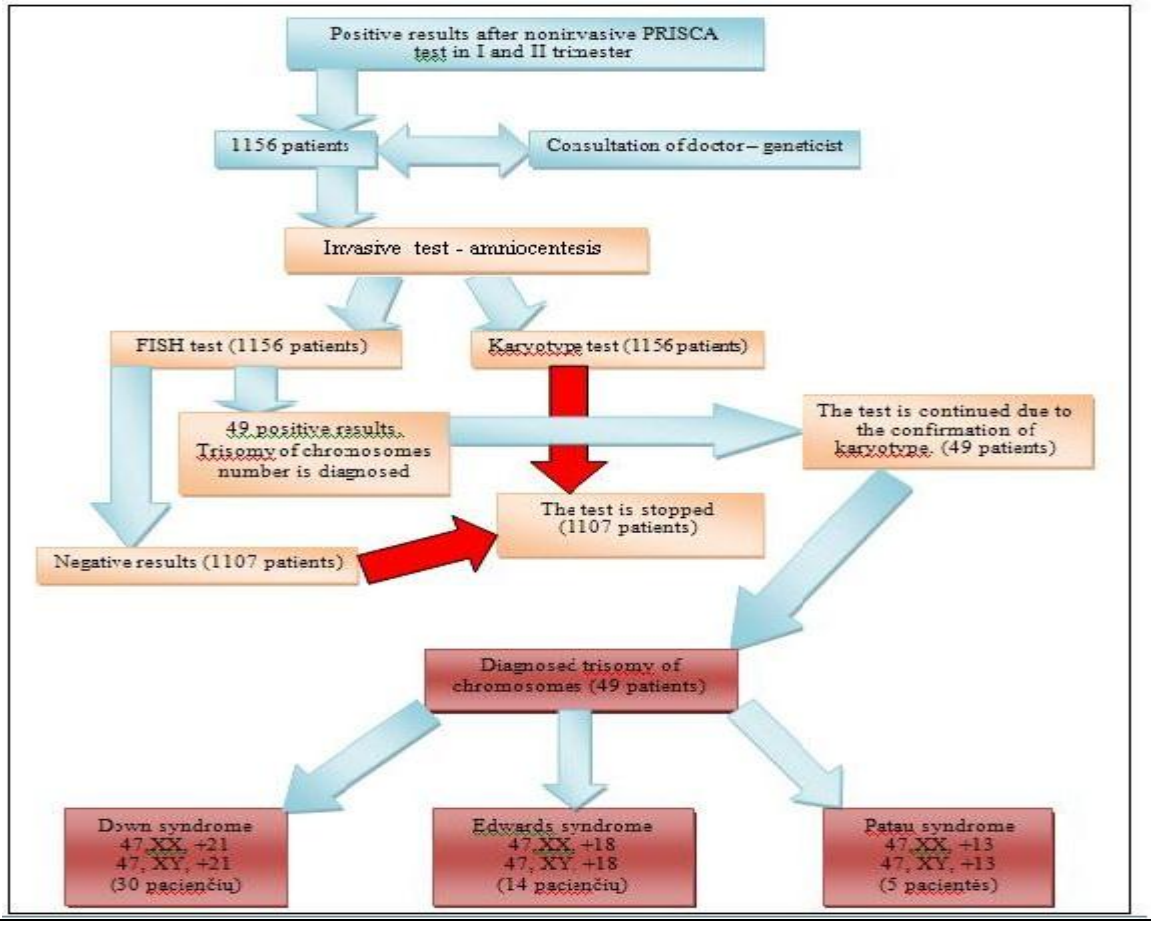
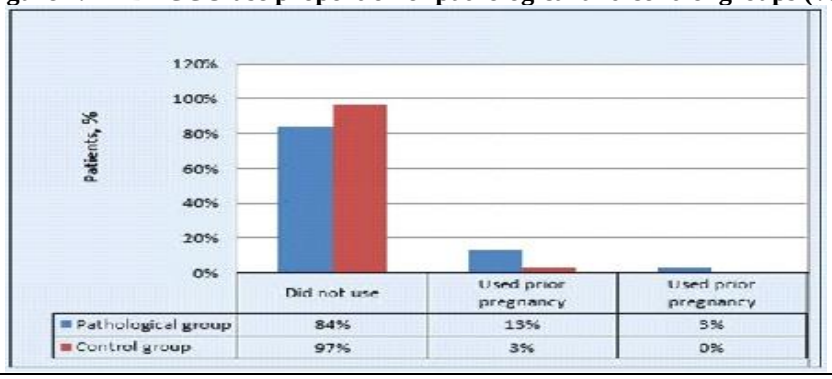


Figure 2. TABACCO use proportion of pathological and control groups (%)



DISCUSSIONS

49 cases of abnormalities of chromosome number was found in 1159 patients in the study material. Of 49 pathological conditions in 30 patients (61%) were diagnosed with an extra 21st chromosome 14 patients (29%) were diagnosed with an additional 18 chromosome, and only 5 patients (10%) were diagnosed with extra chromosomes 13, 19 - 34 patients have been diagnosed with fetal abnormalities in 31.5% of cases, and 35-44 patients were diagnosed even 68.5% of cases of fetal abnormality.

The results from the study corresponds to the data in the literature. Down syndrome is the most frequently diagnosed trisomies of chromosomes (including neonatal frequency of 1: 700), and the most rarely diagnosed is Patau syndrome (incidence of 1: 10,000) [7]. In addition, the results of the research showed that of previous studies that 35-40 year-old women several times higher risk to give birth to a newborn with Down, Edwards and Patau syndrome, compared with younger women results [8-11]. The largest number of patients was 345, of which 14

patients (4.1%) were diagnosed with fetal anomalies in 2009, 2008 237 patients were examined, of which 9 (3.8%) were diagnosed with fetal abnormality, 2010 216 patients were examined and 3, 7% of abnormalities (that is, one anomaly less) were diagnosed. During 2011, 220 patients were examined and 12 pathologies (5.5%) were diagnosed. The least number of patients were tested in 2007 (136 patients), but 6 pathology (4.4%) related to down, Edwards and Patau syndromes were diagnosed.

CONCLUSIONS

After carrying out the cytogenetic and molecular cytogenetic FISH test results of 1,156 patients in the amniotic fluid, 49 (4.2%) pathological cases were diagnosed in the analysis: 13 (0.4%), 18 (1.3%), and 21 (2.6%) trisomies of chromosomes.

Summarizing the results of the questionnaire (according to records on file) and analysing them using statistical package for social sciences (statistical reliability $p < 0.05$) basic risk and environmental factors (from tabacco, genetic diseases, medication use) that affect fetal anomalies development.

ACKNOWLEDGEMENT: NIL

CONFLICT OF INTEREST: NIL

REFERENCES

1. Palermo G, Colombero L, Schattman G, Davis OK, Rosenwaks Z. Ebolution of pregnancies and initial follow-up of newborns delivered after intracytoplasmic sperm injection. *J Am Med Assoc*, 276, 1996, 1893-1897.
2. Sheets K. B, Blythe G. Crissman, Cori D. Feist, Susan L. Sell, Lisa R. Johnson, Kelly C. Donahue, Diane Masser-Frye, Gail S. Brookshire, Amanda M. Carre, Danielle LaGrave, Campbell K. Brasington. Practice Guidelines for Communicating a Prenatal or Postnatal Diagnosis of Down syndrome, *National Society of Genetic Counselors*, 15, 2011, 42 - 60.
3. Wisanto A, Bonduelle M, Camus M, Tournaye H, Magnus M, Liebaers I, Van Steirteghem A, Devroey P. Obstetric outcome of 904 pregnancies after intracytoplasmic sperm injection. *Hum Reprod*, 11, 1996, 121-130.
4. Langlois S, Brock J.A, Wilson R.D, Audibert F, Brock J.A, Carroll J, Cartier L, Gagnon A, Johnson J.A, Langlois S, Macdonald W, Murphy-Kaulbeck L, Okun N, Pastuck M, Senikas V. Current status in non-invasive prenatal detection of down syndrome, trisomy 18, and trisomy 13 using cell-free DNA in maternal plasma. *Obstet gynaecol Canada*, 15, 2013, 81 - 125.
5. Wennerholm UB. Cryopreservation of embryos and oocytes: obstetric outcome and health in children. *Hum Reprod*, 5, 2000, 18-25.
6. Skirton H, Goldsmith L, Jackson L, Lewis C, Chitty L. Offering prenatal diagnostic tests: European guidelines for clinical practice guidelines. *European Journal of Human Genetics*, 22, 2014, 580-586.
7. Momsen G, Sundberg K, Friis-Hansen L, Jorgensen F.S. First-trimester risk calculation for trisomy 13, 18, and 21: comparison of the screening efficiency between 2 locally developed programs and commercial software. *Clinical Cemistry*, 14, 2011, 57 - 72.
8. Wennerholm UB, Albertsson-Wikland K, Bergh C, Hamberger L, Niklasson A, Nilsson L, Thiringer K, Wennergren M, Wikland M, Borres MP. Postnatal growth and health in children born after cryopreservation as embryos. *Lancet*, 351, 1998, 1085-1090.
9. Harada S, Korf B. R. Overview of molecular genetic diagnosis.*Current Protocols in Human Genetics*, University of Alabama at Birmingham, 12, 2006, 29 - 41.
10. Wada I, Macnamee MC, Wick K, Bradfield JM, Brinsden PR. Birth characteristics and perinatal outcome of babies conceived from cryopreserved embryos. *Hum Reprod*, 9, 1994, 543-546.
11. Hill P. Down Syndrome Prevalence In The United States. American Academy of Pediatrics, 11, 2009, 9 - 21.