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FREQUENCY AND STRUCTURE OF CONGENITAL DEVELOPMENT ANOMALIES

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Abstract

To study the frequency and structure of congenital malformations based on the results of prenatal screening (ultrasound, biochemical and cytogenetic screening). Improving perinatal outcomes and lowering the birth rate of children with disabilities. Strengthening preventive work in the field of "Protection of motherhood and childhood."

Key words: *prenatal screening, ultrasound, biochemical screening, cytogenetics, fetus, congenital malformations, disability, clinic, prevention.*

I. Relevance

In order to improve perinatal indicators, the healthcare system in Uzbekistan is being modernized as part of the “Protection of Motherhood and Childhood” program. The introduction of new technologies, international standards and protocols today shows its positive results. However, among the causes of perinatal mortality, congenital malformations (CM) occupy a significant first and second place. These problems indicate that perinatology, embryology, pediatrics, genetics, obstetrics and gynecology, prevention are still relevant issues that have yet to be studied [1,3,5,8].

According to medical genetics advisory reports, in the structure of congenital malformations - congenital heart defects (CHD) take the first place, making up 37%, the second - the musculoskeletal system anomalies (15%), and the third place is occupied by the anomalies of the urinary system; the gastrointestinal tract is in fourth place (8.6%), chromosome disorders - in fifth place (7.6%), maxillofacial malformations - in sixth place with 6.8%, central nervous system and other rare defects - in seventh place, accounting for 3.9%. In the structure of child mortality, perinatal morbidity is in first place (37.4%), CM in second place (31.8%), traumatic and intestinal infections in third place (9.3%) [2,4,6,7] .

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II. The aim of the study

Is to study the cause and structure of congenital developmental anomalies for 2018-2019 in the Samarkand regional center "Screening of mother and child". We conducted a retrospective study of the results of prenatal screening (ultrasound, biochemical and cytogenetic) (n = 59777).

III. Materials and research methods

A retrospective analysis of pregnancy histories was carried out, the history and features of the clinical course of pregnancy were studied, and prenatal screening (PS) methods were studied. Statistical processing of the material was conducted.

In order to study the frequency and structure of CM in the Samarkand regional center for screening mothers and children in 2018-2019 years a retrospective study of the results of prenatal screening (ultrasound, biochemical and cytogenetic) was conducted (n = 59777).

IV. According to the results of the examination

In 2018-2019, prenatal ultrasound screening (PUS), biochemical and cytogenetic screening for 59,777 pregnant women were performed at the Samarkand regional mother and child screening center.

There 7412 women (12.4%) were examined in the first trimester of pregnancy, 47.671 (79.7%) in the second trimester and 4694 (7.9%) in the third trimester. Of these, a total of 1382 (2.3%) pregnant women revealed fetal abnormalities. Moreover, in 237 (0.4%) women, fetal pathology was detected in the first trimester of pregnancy, in 896 (1.5%) were found in the second trimester, and in 249 (0.4%) pregnant women were found in the third trimester. Among the revealed pathologies of the fetus, the proportion of CM was 1282 (2.1%) cases. CM was diagnosed in 204 (0.34%) pregnant women in the first trimester, in 833 (1.4%) in the second trimester and in 245 (0.4%) in the third trimester. Obstetric pathologies (anembryo, undeveloped pregnancy, cystic skidding, various fetal malformations, antenatal fetal death) were found in 100 (0.17%) pregnant women. Of these, 33 (0.05%) cases were diagnosed in the first trimester of pregnancy, in 63 (0.1%) in the second trimester, and in 4 (0.007%) women in the third trimester. In 983 (1.6%) women, various fetal malformations were diagnosed and, as a result of prenatal ultrasound screening (PUS), were aimed at terminating the pregnancy in accordance with medical recommendations. Of these, 855 (87%) women terminated the pregnancy, while in 757 (88.5%) women the pregnancy was terminated due to fetal CM and in 98 (11.5%) cases due to obstetric pathology.

In 2019, a total of 42,130 pregnant women underwent PUS at the Samarkand Regional Center for Maternal and Child Screening. In 744 of them (1.8%), pathologies were revealed both from the side of the mother and from the fetus. Of these, fetal CM were detected in 728 (1.7%) and obstetric pathology in 16 (0.04%) cases (Table 1).

According to the results of PUS, 744 (1.8%) of 42 130 pregnant women were diagnosed with pathology. Obstetric pathology (anembryo, non-developing pregnancy, cystic skid, antenatal fetal death) was observed in 16 (0.04%) women. CM was found in 728 (1.7%). Of these, 462 (63.5%) had mono-CM, and 266 (35.5%) had several CM (table 2).

Among the congenital anomalies, the proportion of anomalies of the central nervous system (CNS) was 169 (23.2%). In the first three months of pregnancy, the pathology of the central nervous system was monotypic in 23 (3.2%) and multiple types of 11 (1.5%), a total of 34 (4.7%) cases. In the second trimester, the mono type was 57 (7.8%), the multiple type was 47 (6.5%) and overall 104 (14.3%) cases. In the third trimester, the mono type was 18 (2.5%), the multiple type was 13 (1.8%), a total of 31 (4.3%) cases. In the third trimester, the mono type was 18 (2.5%), the multiple type was 13 (1.8%), a total of 31 (4.3%) cases. The PUS was performed by 169 (23.3%) pregnant women, while 98 women diagnosed various pathological changes in the central nervous system of the fetus in the form of mono (13.5%) and multiple types in 71 (9.8%) cases.

The total number of CM of the maxillofacial system was 33 (4.5%). Of these, anomalies were not detected in the first trimester of pregnancy, in the second trimester - mono type 8 (1.1%) and multiple type 15 (2.1%), in only 23 (3.2%) cases. In the third trimester, the mono type was in 7 (0.96%), the multiple type was in 3 (0.4%), for a total of 10 (1.4%) cases. Of all congenital malformations of the maxillofacial system, 15 monotypes (2.1%) and 18 multiple species (2.5%), a total of 33 (4.5%) cases.

Table 1.

Results of prenatal ultrasound screening n = 42130, (in percentages)

Systems	**I-trimester**	**II-trimester**	**III - trimester**	**Total**
Pathology of the fetus 744 (1,8)	118 (15,8)	467 (62,7)	159 (21,4)	744 (100)
CM 728 (1,7)	118 (16,2)	452 (62,1)	158 (21,7)	728 (100)
Mono CM 462 (63,5)	80 (11)	264 (36,3)	118 (16,2)	462 (63,5)
Multiple CM 266 (35,5)	38 (5,2)	188 (25,8)	40 (5,5)	266 (36,5)
Obstetric pathology		15 (2)	1 (0,14)	16 (2,2)
CM of the central nervous system	34 (4,7)	104 (14,3)	31 (4,3)	169 (23,2)
Mono types	23 (3,2)	57 (7,8)	18 (2,5)	98 (13,5)
Multiple types	11 (1,5)	47 (6,5)	13 (1,8)	71 (9,8)
CM of the maxillofacial system		23 (3,2)	10 (1,4)	33 (4,5)

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Mono types	-	8 (1,!)	7 (0,96)	15 (2,1)
Multiple types	-	15 (2,1)	3 (0,4)	18 (2,5)
CM of the spine	12 (1,6)	25 (3,4)		37 (5,1)
Mono types	3 (0,4)	8 (1,1)	-	11 (1,5)
Multiple types	9 (1,2)	17 (2,3)	-	26 (3,6)
CM of the urogenital system	15 (2,1)	90 (12,4)	41 (5,6)	146 (20,1)
Mono types	15 (2,1)	65 (8,9)	38 (5,2)	118 (16,2)
Multiple types	-	25 (3,4)	3 (0,4)	28 (3,8)
CM of the respiratory organs	5 (0,7)	35 (4,8)	12 (1,6)	52 (7,1)
Mono types	1 (0,14)	20 (2,7)	3 (0,4)	24 (3,3)
Multiple types	4 (0,5)	15 (2,1)	9 (1,2)	28 (3,8)
CM of the musculoskeletal system	2 (0,3)	44 (6)	17 (2,3)	63 (8,7)
Mono types	2 (0,3)	28 (3,8)	15 (2,1)	45 (6,2)
Multiple types	-	16 (2,2)	2 (0,3)	18 (2,5)
CM of the digestive organs	18 (2,5)	43 (5,9)	21 (2,9)	82 (11,3)
Mono types	8 (1,1)	22 (3)	17 (2,3)	47 (6,5)
Multiple types	10 (1,4)	21 (2,9)	4 (0,5)	35 (4,8)
CM of the circulatory system	6 (0,8)	66 (9,1)	24 (3,3)	96 (13,2)
Mono types	2 (0,3)	34 (4,7)	18 (2,5)	54 (7,4)
Multiple types	4 (0,5)	32 (4,4)	6 (0,8)	42 (5,8)

Other CM	26 (3,6)	22 (3)	2 (0,3)	50 (6,9)
Obstetric pathology		15 (2,1)	1 (0,14)	16 (2,2)

In total, VAR was diagnosed in 63 (8.7%) cases, of which a monotype in 2 (0.3%) cases in the first trimester of pregnancy. Multiple type is was not detected; In the second trimester - mono type 28 (3.8%) and multiple type 16 (2.2%), a total of 44 (6%); in the third trimester, monotype 15 (2.1%) and multiple type 2 (0.3%) were found, in total 17 (2.3%) cases. In total, 63 (8.7%) cases were identified, of which 45 (6.2%) were monotypes and 18 (2.5%) were multiple types of CM.

The total number of CM of the spine was 37 (5.1%), which was diagnosed in all three trimesters of pregnancy as follows: mono type 3 (0.4%), multiple type 9 (1.2%) in the first trimester, overall 12 (1 , 6%); In the second trimester, monotype 8 (1.1%) and the multiple type 17 (2.3%) were present, overall 25 (3.4%). No such abnormalities were found in the third trimester of pregnancy. Of all congenital abnormalities of the spinal system, 11 monotypes (1.5%) and multiple types 26 (3.6%), a total of 37 (5.1%) cases.

CM of the urogenital system was detected in 146 (20.1%) cases, of which 15 (2.1%) were of the same type in the first trimester, and 15 (2.1%) were absent; In the second trimester, monotype was 65 (8.9%), and the multiple type was 25 (3.4%), overall 90 (12.4%). During the third trimester of pregnancy, monotype 38 (5.2%) and multiple type 3 (0.4%) were found, altogether 41 (5.6%). The total number of monotypes of congenital anomalies of the genitourinary system was 118 (16.2%), and the number of multiple types was 28 (3.8%), a total of 146 (20.1%) cases.

In total there were 52 (7.1%) CM of the respiratory system: mono type 1 (0.14%), multiple type 4 (0.5%), altogether 5 (0.7%) in the first trimester of pregnancy; In the second trimester - mono type 20 (2.7%) and multiple type 15 (2.1%), a total of 36 (4.8%); In the third trimester, mono type 3 (0.4%) and multiple type 9 (1.2%) were found, overall 12 (1.6%). Twenty four (3.3%) monotypes of congenital malformations of the respiratory system and 28 (3.8%) multiple types were identified, accounting for 52 (7.1%) cases.

A total of 82 (11.3%) CM of the digestive organs were recorded: in the first three months, monotype 8 (1.1%), multiple type 10 (1.4%), overall 18 (2.5%); In the second trimester, mono type 22 (3%) and multiple type 21 (2.9%), a total of 43 (5.9%); In the third trimester, monotype 17 (2.3%) and multiple type 4 (0.5%) were found, altogether 21 (2.9%). A total of 47 (6.5%) monotypes and 35 (4.8%) multiple types of CM of the digestive organs cases were identified.

CM of the circulatory system were diagnosed in only 96 (13.2%) cases: mono type 2 (0.3%), multiple type 4 (0.5%), overall 6 (0.8%) in the first three months; In the second trimester, mono type 34 (4.7%) and multiple type 32 (4.4%), a total of 66 (9.1%); In the third trimester, monotype 18 (2.5%) and multiple type 6 (0.8%) were found, altogether 24 (3.3%). Of all CM of the circulatory system, 54 monotypes (7.4%) and 42 multiple types (5.8%) were found, altogether it was 96 (13.2%).

Other types of CM were 50 (6.9%): non-immune accumulation of water in the extravascular spaces of the fetal body, 1 (2%) in the first trimester, 4 (8%) in the second trimester, overall 5 (10%). Not detected in the third trimester of pregnancy. Multicellular cystic fetal hygroma with accumulation of non-immune water in the extracellular spaces of the fetal body was present in 25 (50%) in the first trimester, in 16 (32%) in the second trimester, and in 2 (4%) in the third trimester, a total of 43 (86%) cases. The fetal neck zone was detected in 2 (4%) individuals with multicellular cystic hygroma, ascites, and multiple fluid (table 2).

Table 2

The coefficient of comparison of the incidence and structure of CM with other countries (in%)

Systems	**Yakutia**	**Russian Federation**	**EUROCAT**	**Samarkand regional center of "Screening of mother and child"**
The cardiovascular system	37,0	33,89	20,63	13,2
Musculoskeletal system	15,0	19,77	24,84	13,8
Urogenital system	13,0	17,18	15,17	20,1
Digestive system	8,6	8,16	12,78	11,3
Central nervous system	3,9	6,53	12,61	23,2
Other	3,8	5,92	-	6,9
Chromosomal	7,6	4,99	10,38	3,55
Face system	6,8	1, 53	-	4,5
Respiratory system	1,8	0,70	-	7,1

The frequency of occurrence of numerical autosomal abnormalities in prenatal biochemical studies was also studied. During the first trimester of pregnancy, 3666 pregnant women underwent PAPP-A / b-HCG, a prenatal biochemical screening of double signs. In 1512 women, the placental condition factor was studied. Interpretation of the results, the individual risk of Down syndrome was 1/100 and higher (0.85%) in 31 people. At 40, the risk was 1 / 101-1 / 360 (1.1%). In 3595 people, the risk was very 1: 1001 and low (98.1%). At 60, the individual risk of Edward's syndrome was 1/101 low (1.6%), with no high risk.

In the second trimester of pregnancy in 2018, 3276 pregnant women underwent prenatal biochemical screening for AFP / XG and 84 for unconjugated estriol. According to the results of the study: the

individual risk of Down syndrome was 1/100 or more in 14 people (0.4%), in the range 1 / 101-1 / 360 (5.7%) in 188 people and 1 / 361-1 / 1000 in 222 people (6.8%), 2852 were found 1: 1001 and the risk was low (87.1%). Studies on Edward's syndrome have not been conducted. The individual risk of neural tube defects was low (99.6%) in 3262 and high (0.4%) in 14 cases.

According to 2019 data, prenatal biochemical screening was performed in the second trimester of pregnancy: AFP / XG in 8191 pregnant women, conducted according to conjugated estriol. The results were as follows: Individual risk of Down Syndrome was 1/100 or more in 1.1 (1.6%), 1 / 101-1 / 360 (4%) in 330, and 1 / 361-1 / 1000 (2.4) in 198, respectively. 7295 were found to be 1: 1001 or less (88.6%). It was found that the individual risk of Edward's syndrome is 1/100 and higher (83%) in 6796 and 1/101 and lower (1.3%) in 103, respectively. The individual risk of neural tube defects was low in 6960 (85%) and high in 26 (0.3%).

Individual risk of Edward's syndrome was high. Edward's syndrome is a multiple defect, so the risk is high. Symptoms of Edwards syndrome: the birth of a child with a low weight of 2 kg, 100 g - 2 kg, 200 g; irregular shape of the face - the head is small, dolichocephalic, that is, extending to the neck; gidrotsefaliya; fingers or toes are partially and completely attached to each other; signs of “cleft palate and cleft lip”, deformation of the face: narrow forehead, wide neck, lower chin, undersized mouth, small, short mouth, high palate, irregular teeth; narrow eyelids, deep eyebrows, narrow nose; the neck is short, the skin of the neck area forms a fold; ears are improperly shaved, branches are small or absent, horizontally elongated and the ear canals are narrow. Exographic signs of Edwards syndrome, umbilical artery agenesis, small placenta, excessive moisture, and the detection of multiple defects. Data for 2018-2019 almost equivalent to each other.

The results of cytogenetic testing, in 2019 121 tests were performed. Of these, 103 (85.1%) were performed in children under 14 years of age, 8 (6.6%) in children aged 14-18 and 10 (8.3%) in adults. In 23 of them (19%), the karyotype was normal. Down syndrome was detected in 78 (64.5%), of which 70 (57.8%) had the classic form of Down syndrome (complete trisomy), 6 (5%) had a mosaic form Down syndrome , and 2 (1.7%) a translational form of Down Syndrome. Also, 6 (5%) Shereshevsky-Turner syndrome, 2 (1.7%) Kleinfelter syndrome, 1 (0.8%) other chromosomal pathologies and 11 (9.1%) signs of fetal malformation were indentified.

V. Conclusion

Among CM, the first place is occupied by anomalies from the side of the central nervous system (23.2%), the second is the urogenital system (20.1%), the third is the musculoskeletal system (13.8%), the fourth is cardio-vascular system (13.2%%), in fifth place - the digestive system system (11.3%), the sixth is the respiratory system (7.1), other anomalies in seventh place (6.9%), the eighth amounted to the maxillofacial system (4.5%) and ninth place - chromosomal diseases (3.55%).

The risk of Edwards syndrome, PAPP-A / b-HCG in the first trimester of pregnancy, prenatal biochemical screening with two symptoms and placental growth factor was low - 1.6%, a high risk was not detected Interpretation of the results showed that the low risk of Edwards syndrome in prenatal biochemical screening ranged from 1.3% to 1.6%. High risk was detected only in the second trimester of pregnancy in 83% of cases.

The individual risks of Down Syndrome were PAPP-A / b-HCG, prenatal biochemical screening of double risk and a high risk of developing placental growth factor - 0.85%, medium risk - 1.1% and low risk - 98.1%. , According to the results of prenatal biochemical screening AFP / XG, dual trait and unconjugated estriol in the second half of pregnancy, the risk of Down syndrome is 1.6%, relatively high risk - 4%, moderate risk - 2.4% and low risk - 88.6 % Interpretation of the results showed that the risk of Down syndrome during prenatal biochemical screening was 0.85-1.6%, and the relative high risk was found only in the second half of pregnancy and amounted to 4%. The average risk was 1.1-2.4%, and the low - 88.6-98.1%.

Based on the results of prenatal biochemical studies, it was found that the risk of total preeclampsia is 27.6%, and the risk of individual neural tube defects was low - 85% and high - 0.3%.

According to the interpretation of the results of cytogenetic examination, the norm of the karyotype was revealed in 19% of the examined. Down syndrome was detected in 64.5%, Shereshevsky-Turner syndrome in 5%, Klinefelter syndrome in 1.7%, other chromosomal pathologies in 0.8%, and 9.1% had fetal tumor clinics.

Recommendations

To reduce the number of births with VAR and obstetric pathologies, it is advisable to do the following:

1. Follow a healthy lifestyle, follow a healthy diet rich in minerals and vitamins.

2. Get genetic counseling.

3. Conducting periconceptive prophylaxis.

4. Qualitatively and effectively conduct classes of the "Mother School". To be able to involve pregnant women and their partners who are preparing for the birth process to this trainings.

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