International Journal of Science and Research (IJSR)
ISSN: 2319-7064
ResearchGate Impact Factor (2018): 0.28 | SJIF (2019): 7.583

Profile of Pregnancy undergoing Amniocentesis in the Sanglah Birth Defect Integrated Center (SIDIC) Fetal Maternal Outpatient Department in Sanglah Hospital: A Descriptive Study from June 2017-June 2019

Lydia Linasari Sinarta[1], I Wayan Artana Putra[2]

[1]Obstetrics and Gynecology Department, Medical Faculty of Udayana University / Sanglah Hospital

[2]Fetal Maternal Division, Obstetrics and Gynecology Department, Medical Faculty of Udayana University / Sanglah Hospital

Abstract: ***Objective: Amniocentesis is useful for detecting chromosomal abnormalities, neural tube defects, and genetic disorders. Sanglah Hospital is one of the hospitals implementing surveillance for congenital abnormalities through the Sanglah Integrated Birth Defect Center (SIDIC) program. The purpose of this study was to determine the profile of pregnancies undergoing amniocentesis procedures in the SIDIC Fetal Maternal Outpatient Department in Sanglah Hospital from June 2017 to June 2019. Method: This is a study using the secondary data sources from medical records of patients in the period of June 2017 to June 2019 at SIDIC Sanglah Hospital Denpasar. Results: During the period June 2017-June 2019, there were 32 patients underwent amniocentesis. The result was that there were 7 patients (22%) with chromosomal abnormalities and 25 patients (78%) had no chromosomal abnormalities. Seven patients with chromosomal abnormalities were all in a correlation between amniocentesis, ultrasound, and abnormalities found at birth. Trisomy 18 is the most chromosomal abnormality found in 4 cases, the remaining trisomy 23, tetraploid and p13p15 deletions 1 case each. Outcomes of baby with chromosomal abnormalities were 4 died (57.14%), 2 alive (28.57%), and 1 was terminated (14.28%). In 25 patients without chromosomal abnormalities, 23 patients were found to have abnormalities on ultrasound and congenital abnormalities at the time of birth, while in 2 more patients only found abnormalities on ultrasound. Outcomes of the baby without chromosomal abnormalities were 9 died (36%), 15 alive (60%) and 1 was terminated (4%). Head or CNS abnormalities and the cardiovascular system are the most common types of congenital abnormalities based on the most common ultrasound examination found. Hydrocephalus, labiopalatoschizis, and omphalocele are major congenital abnormalities most often detected at birth. Conclusion: During the period June 2017 - June 2019 there were 32 patients undergoing amniocentesis at the SIDIC Fetal Maternal Outpatient Department in Sanglah Hospital. All chromosomal abnormalities found in amniocentesis correlate with ultrasound abnormalities and abnormalities found at birth***

Keywords: amniocentesis, congenital abnormalities, SIDIC

1. Introduction

A birth defect or congenital anomalies are developmental abnormalities that occur during pregnancy or the prenatal period.[1] Congenital abnormalities can be caused by single-gene defects chromosomal abnormalities, multifactorial inheritance factors, teratogens, and micronutrient deficiencies.[2,3] The most common congenital abnormalities in Indonesia are talipes equinovanus, orofacial cleft, neural tube defects, abdominal wall defects, atresia ani, hypospadias or epispadias, conjoined twins, and microcephaly.[4]

Congenital abnormalities are a global health problem and an important cause of mortality and morbidity in children. It is estimated that each year around 3% of babies are born with birth defects.[5] Surveillance studies in the population of newborns in the United States show the prevalence of birth defects is 29.2 in 1,000 live births.[6] In developing countries, the estimated total prevalence of birth defects according to WHO is 47.2 in 1,000 live births. In developing countries, the estimated total prevalence of birth defects according to WHO is 47.2 in 1,000 live births. In Southeast Asia, the prevalence of birth defects is 7%. The prevalence of birth defects in Indonesia also reaches 59.3 per 1,000 birth defects.[7] Baby mortality due to congenital abnormalities occupies the fourth highest proportion of causes of baby mortality in 2015 that is 303,000 and is the second-highest cause of baby death based on Riskesdas 2007 data.[4]

Prenatal surveillance and diagnosis of congenital anomalies are very important. Amniocentesis is the most common procedure used for prenatal diagnosis. Amnio- centesis is useful for detecting chromosomal abnormalities, neural tube defects, and genetic disorders. Abnormalities encountered such as trisomy 21, spina bifida, and cystic fibrosis are definitively diagnosed through amniocentesis.[8] Prenatal diagnosis, and screening has long been carried out in developed countries.[9,10] In America, an amniocentesis procedure is carried out up to 200,000 times in a year.[11] However, due to the lack of available data and resources, prenatal diagnoses are not commonly done in middle to lower-income countries.[12] Meanwhile in Indonesia, a congenital abnormality surveillance program is being held, in which Sanglah Hospital is one of the implementing hospitals.[4]

Therefore, research conducted at the Sanglah Integrated Birth Defect Center (SIDIC) Fetal Maternal Outpatient Department in Sanglah Hospital has contributed to improving data related to congenital abnormalities in Indonesia by obtaining data on the incidence of congenital abnormalities and pregnancy profiles, specifically in the group of patients undergoing amniocentesis since amniocentesis was first carried out on June 2017.

2. Methods

This study was a descriptive retrospective study conducted at *Sanglah Birth Defect Integrated Centre* (SIDIC) Fetal Maternal Outpatient Department in Sanglah Hospital Denpasar, from June 2017 to June 2019. The study sample was taken from the medical records of all women diagnosed with congenital abnormalities of the fetus undergoing amniocentesis procedures who came to Sanglah Birth Defect Integrated Centre Fetal Maternal Outpatient Department Sanglah Hospital Denpasar on June 2017 – June 2019. The target population is pregnant women with a diagnosis of congenital abnormalities in the fetus Ethical approval and research permission were obtained from the hospital ethics committee of the Medical Faculty of Udayana University

The data obtained is then collected, processed, and presented in table and narrative form. Data taken from the medical record is the number of incidents based on amniocentesis examination and the distribution of birth defects. Besides, the data is then analyzed descriptively based on gestational age, parental age, parental education level, history of maternal parity and abortion, identification of risk of disease in the first-degree offspring, exposure to drugs during preconception and pregnancy, and consumption of folic acid.

3. Results

The results showed patients undergoing amniocentesis in the final semester of 2017 there were 4 patients, in 2018 there were 16 patients, and in the early semester of 2019, there were 12 patients. A total of 32 patients, with 7 patients (21,875%) detected having chromosomal abnormalities and 25 patients (78,125%) did not have chromosomal abnormalities. Total labor during the period June 2017 - June 2019 at Sanglah Hospital was 2273 childbirth so that if counted there were 7 births (0.31%) with chromosomal abnormalities and 25 births (1.09%) with congenital abnormalities without chromosomal abnormalities.

The mean age of parents of the study subjects was over 30 years, with the mean age of the mother being 31.8 years and the mean age of the father was 34.3 years. The mean gestational age in this study was 26.75 weeks with the majority in trimester 2 and trimester 3 (50% and 46.8%). The result of maternal and paternal age subjects, ethnicity, and parity are shown in Table 1. The results of karyotyping after amniocentesis are shown in Table 2. Also, results showed that the majority of study subjects (25 subjects) did not find any chromosomal abnormalities (78.125%) and trisomy 18 or Edward syndrome is the most chromosomal abnormality detected.

Ultrasound examination is one of the non-invasive prenatal screening methods in detecting birth defects. Ultrasound examination in this study revealed a variety of abnormalities found in research subjects who performed amniocentesis procedures at Sanglah Hospital Denpasar. Based on the ultrasound examination, found 7 patients with multiple congenital abnormalities, and 25 patients with a single disorder. Head or CNS, cardiovascular system abnormalities, and gastrointestinal system abnormalities are the most common types of congenital abnormalities based on the most frequent ultrasound examinations are shown in Table 3. Whereas if based on the type of disease, the most abnormalities based on the results of an ultrasound examination are omphalocele.

Table 1: Characteristics of the participants

Characteristics	Mean	Total (n=32)	Percentage (%)
Mother's Age			
≤20 years old		1	3.1%
21-25 years old		3	9.4%
26-30 years old	31.8 years old	13	40.6%
31-35 years old		7	21.9%
36-40 years old		2	6.2%
≥40 years old		6	18.8%
Father's Age			
≤20 years old		0	0%
21-25 years old		3	9.4%
26-30 years old	34.3 years old	10	31.2%
31-35 years old		7	21.9%
36-40 years old		4	12.5%
≥40 years old		8	25%
Ethnic			
Java		3	9.4%
Bali		28	87.5%
Lombok		1	3.1%
Parity			
0		10	31.25%
1	1.25	8	25%
2		10	31.25%
3		4	12.5%
Gestational Age			
1st Trimester	26.75 weeks	1	3.1%
2nd Trimester		16	50%
3rd trimester		15	46.8%

Table 2: Post amniocentesis karyotyping results

Characteristics	**Total *(n=32)***	**Percentage (%)**
Without chromosomal abnormalities	**25**	**78.125%**
46, XX	11	34.375%
46, XY	14	43.75%
With chromosomal abnormalities	**7**	**21.875%**
Trisomi 13	1	3.125%
Trisomi 18	4	12.5%
Tetraploid	1	3.125%
Delesi p13p15	1	3.125%

Table 3: Types of major congenital abnormalities based on ultrasound examination

Characteristics	N
Polyhydramnios	5
Head abnormalities	9
Ventriculomegaly	2
Frontal browsing	1
Vein of Galen Aneurysm, Mega Fossa Anterior	1
Hypogenesis of Corpus Callosum	2
Lobar Holoprosencephaly, Microcephaly	1
Hydrocephalus	1
Thickening of the nuchal cord	1
Facial abnormalities	2
Labioschizis	1
Labiognatopalatoschizis	1
Gastrointestinal system abnormalities	8
Omphalocele	4
Gastroschisis	1
Esophageal atresia	3
Ascites	3
Genitourinary system abnormalities	3
LUTO (Posterior urethral valve), bilateral hydronephrosis	1
Pyelectasis renal dextra	1
Multicystic dysplastic left kidney (Potter Type II)	1
Cardiovascular system abnormalities	9
Single umbilical artery	1
Fetal sustained bradycardia, fetal hydrops	1
Ventricular septal defect	2
Dextroposition of the heart	2
Cardiomegaly	2
Atrial septal defect	1
Respiratory system abnormalities	1
Diaphragmatic hernia	1
Extremities and musculoskeletal abnormalities	2
Short femur and humerus	1
Thanatophoric	1

Table 4 shown the types of congenital abnormalities detected at birth. Hydrocephalus and labiopalatoschisis are major congenital disorders that are most often detected at birth.

Table 4: Congenital anomalies detected at birth

Characteristics	**N**
Head abnormalities	
Hydrocephalus	3
Spina bifida	1
Microcephaly	1
Facial abnormalities	
Labiopalatoschizis	3
Labiognatopalatoschizis	2
Micrognatia	1
Gastrointestinal system abnormalities	
Omphalocele	3
Gastroschisis	1
Esophageal atresia	1
Ascites	1
Genitourinary system abnormalities	
LUTO (Posterior urethral valve), bilateral hydronephrosis	1
Cardiovascular system abnormalities	
Ventricular septal defect (VSD)	1
Dextroposition of the heart	1
Atrial septal defect (ASD)	1
Cyanotic congenital heart disease	1
Tetralogy of Fallot (TOF)	1
Patent ductus arteriosus (PAD)	1
Ebstein anomaly	1
Respiratory system abnormalities	
Diaphragmatic hernia	1
Extremities and musculoskeletal abnormalities	
Thanatophoric	2
CTEV	1

In this study obtained from 32 patients, only 7 patients were judged to have a similarity between abnormalities in chromosome analysis, findings on ultrasound, and congenital abnormalities at birth (table 5). Seven chromosomal abnormalities have postpartum congenital abnormalities in the form of four trisomy abnormality 18, one trisomy abnormality 13, one tetraploid abnormality, and one p13p15 deletion abnormality. In tetraploid abnormality, there are children with short femurs and short humerus. The profile of mothers with a tetraploid abnormality is 33 years old with 1 parity. In trisomy 18 abnormality, four different profiles of mothers were found, 32 years old with 1 parity, 38 years old with 2 parity, 41 years old with 3 parity, up to 42 years old with 3 parity. In p13p15 deletion abnormality with 35 years old mother with 2 parity. In trisomy abnormality 13 with 34 years old mother's age profile with 1 parity. Also, table 6 shown the similarities between amniocentesis, ultrasound, and birth abnormalities.

Table 5: Profile of Amniocentesis Examination, Ultrasound, and Birth Abnormalities

Gestational age	Chromosome abnormalities	Ultrasound examination	Postnatal Congenital Abnormality	Baby out Come
31 weeks	Tetraploid	The short femur, short humerus	Same	Termi-nation
26 weeks	Trisomy 13	Labiognatopalatoschiziz, VSD, dextroposition of the heart diaphragmatic hernia	Same	Died
20 weeks	Trisomy 18	Gastrointestinal system disorders	Same	Alive
21 weeks	Trisomy 18	Labiopalatoschisis, arthrogryposis	Same	Died
27 weeks	Trisomi 18	CTEV, micrognatia	Same	Died
28 weeks	Trisomy 18	Cardiomegaly	Same	Died
20 weeks	Delesi p13p15	Omphalocele	Same	Alive

Table 6: The similarity between amniocentesis, ultrasound, and birth abnormalities

Characteristic	n	%
Ultrasound abnormalities, with chromosomal abnormalities, and with congenital abnormalities	7	21.90%
Ultrasound abnormalities, without chromosomal abnormalities, and with congenital abnormalities	23	71.90%
Ultrasound abnormalities, without chromosomal abnormalities, and congenital abnormalities	2	6.20%

Overall baby outcomes can be seen in Table 7, where 17 alive (53.1%), 13 died (40.6%), and 2 were terminated (6.3%). Of the 13 babies who died the majority had congenital abnormalities in the cardiovascular system (61.53%). Table 8 shows from a total of 25 babies born without chromosomal abnormalities, 15 alive (60%), 9 died (36%), and 1 was terminated (4%). Of the 15 live babies, there are 2 babies without chromosomal abnormalities and congenital abnormalities, while 9 of the babies who died all have congenital abnormalities. The gender of 32 babies, obtained as many as 16 male babies (50%), 14 female babies (43.8%), and termination of 2 babies (6.2%) are shown in Table 9.

Table 7: Overall baby outcome

Characteristic	N	%
Alive	17	53.10%
Termination	2	6.30%
Died	13	40.60%

Table 8: The outcome of Babies Without Chromosomal Abnormalities

Characteristic	N	%
Alive	15	60%
Termination	1	4%
Died	9	36%

Table 9: Gender of baby

Characteristic	N	%
Alive	15	60%
Termination	1	4%
Died	9	36%

4. Discussion

In this study, it was found The mean age of parents of the study subjects was over 30 years, with the mean age of the mother being 31.8 years and the mean age of the father was 34.3 years (table 1). Other studies assessing congenital anomaly abnormalities in Saudi Arabia have similar results: the average maternal age is 29 years and the average gestational age at diagnosis is 30 weeks.[13] Another study

said increasing maternal age (>35 years old) reduced the risk of major congenital abnormalities until the age of 40 years after which the risk has increased again (RR = 0.66, 95% CI 0.59-0.74, p <0.001). Whereas age> 40 years is also at risk of 1.4 times (95% CI 1.1-1.8).[14] This is quite different from various studies linking congenital defects with increased maternal and paternal ages. Based on research from Zhang et al., The risk of chromosomal abnormalities increases in old maternal age. The risk of chromosomal abnormalities increased 6.64 times (95% CI 5.55-7.93), the risk for trisomy 21 increased 6.83 times (95% CI 5.63-8.3), and the risk for trisomy 18 increased 4.06 times (95% CI 2.09-7.9) in mothers aged over 35 years compared to women aged 25-29 years.[15] The difference between this study and other studies cannot be concluded because this study does not include populations without birth defects so that their relative risks cannot be calculated. This study only assessed populations at risk of birth defects undergoing amniocentesis examination.

The mean gestational age in this study was 26.75 weeks with the most majorities in trimester 2 and trimester 3 (50% and 46.8%) are shown in Table 1. According to the guidelines, amniocentesis examinations were carried out by 15-20 weeks or in trimester 2. A meta-analysis study concluded that amniocentesis in early pregnancy compared to the second trimester would increase the risk of intrauterine infant death (IUFD) (7.6% versus 5.9%). Other risks include spontaneous abortion, congenital anomalies such as talipes.[16]

Based on the results of chromosome analysis, the majority of study subjects (25 subjects) did not find any chromosomal abnormalities (78.125%). Only 21,875% of the subjects detected had chromosomal abnormalities. The percentage of abnormalities based on chromosome analysis can be seen in Table 2. A congenital anomaly surveillance study in Europe states that of all cases of chromosomal abnormalities, almost 75% of the causes are trisomy 21, 18, and 13. The highest prevalence of trisomy is trisomy 21 which is 23 in 10,000 births, trisomy 18 is 5.9 in 10,000 births, and trisomy 13 is 2.3 in 10,000 births.[17] In this study, chromosomal abnormalities with the highest incidence were caused by trisomy 18. No karyotyping results obtained from amniocentesis in the form of trisomy 21, although this trisomy is most often found. This may be due to the lack of early USG's ability to detect trisomy 21 signs on early trimester antenatal examination (nuchal translucency> 3mm, flat nasal bridge) so that it is missed and the patient is not referred for amniocentesis.

Ultrasound examination in this study revealed a variety of abnormalities found in research subjects who performed the amniocentesis procedure at Sanglah Hospital Denpasar. Based on the ultrasound examination showed in table 3, there were 7 patients with multiple congenital abnormalities and 25 patients with a single disorder. Most types of abnormalities detected are head abnormalities, cardiovascular system abnormalities, and gastrointestinal system abnormalities. Based on research from Sallout et al who assessed ultrasound of 30,632 cases and found major congenital anomaly abnormalities of 5.21%. The study found that most abnormalities were the central nervous system and the genitourinary system.[13] This is in correlates with research from Onyambu et al in Kenya who concluded that head abnormalities were the most congenital anomaly found from USG examination.[18] This is possible because the central nervous system abnormalities can be assessed from the first trimester, especially in extreme cases such as anencephaly. To determine the existence of central nervous system abnormalities through ultrasound assessment of three directions, namely thalamic view, ventricular view, and cerebellar view.[19]

Besides, the sensitivity of ultrasound examination for assessment of congenital anomalies of the central nervous system is also best. From a study found sensitivity reached 88%, the same as sensitivity to anomalies in the urinary system. whereas for the sensitivity of gastrointestinal abnormalities is 54%, musculoskeletal disorders with a sensitivity of 37%, cardiovascular abnormalities are 28% sensitivity, and the lowest is cleft lip and palate with a sensitivity of only 18%.[20]

In this study, there were seven chromosomal abnormalities with postpartum congenital abnormalities in the form of four trisomy abnormalities 18, one trisomy abnormality 13, one tetraploid abnormality, and one p13p15 deletion abnormality are shown in Table 5. Of the seven profiles of mothers with infants of chromosomal abnormalities and postpartum congenital abnormalities, it was concluded that all mothers were over 30 years old with a mean age of 36 years. In studies in Australia and the United Kingdom, it was found that the incidence of trisomy 13 and 18 increases in pregnancies with mothers over 35 years old.[21,22] This is different in several studies which state that maternal age is not related to polyploid abnormalities, only to aneuploid disorders. Although the study found a mean age of mothers with a baby with a chromosome abnormality in the form of a tetraploid are 31 years, the results of the analysis of the relationship were not found to be statistically significant.[23] From a study of congenital disorders based on reports in Europe also found no statistically significant relationship between maternal age and deletion disorders.[17]

The number of parities of mothers with infants with chromosomal abnormalities and other congenital disorders in this study varied between subjects, namely the first three parity, two-second parity, and two-third parity. The Duong et al study concludes that nullipara is associated with an increased risk of limb disorders, omphalocele, diaphragmatic hernias, gastroschisis, cardiac septal defects, Tetralogy of Fallot, hypospadias, esophageal atresia, and hydrocephalus. In this study, it was found that nullipara mothers had children with limb abnormalities, one gastrointestinal abnormality, and one with diaphragmatic hernias in which all three were following the results of the Duong et al.[24] Whereas multiparity is known to be associated with cardiovascular, urogenital, and gastrointestinal system abnormalities,[25] wherein this study found one subject of cardiomegaly in mothers with second parity and one subject with omphalocele in mothers with second parity.

Based on observational studies of amniocentesis and ultrasound examination of patients with birth defects found only 2 babies with live outcomes. It is known that trisomy 13

(Patau's syndrome) and trisomy 18 (Edward's syndrome) are fatal genetic disorders with a prevalence of live births of only 1.4-2 births in 10,000 births.[26] Besides, the risk of death of baby with genetic disorders is also very high, with only 1 baby out of 12 surviving up to> 1 year of age with an average life span of 2 weeks.[27] In this observational study, only 1 trisomy 18 infants were found with the live outcome. Trisomy 13 is a fatal genetic disorder, about 50% of babies with trisomy 18 can survive longer than 1 week and 5-10% of babies can survive past the first year. Compared with trisomy, major defect defects are said if there are abnormalities of the cardiovascular system and the central nervous system and craniofacial. The risk of death is also related to the prematurity and birth weight of babies.[28] This allows an explanation of differences in outcomes of babies with the same trisomy.

Deletions in the European register study found a prevalence of 1.86 in 10,000 births. While the prevalence of microdeletions was found to be 0.96 in 10,000 births. The number is less than the prevalence of trisomy 18 which is 5.9 in 10,000 births and trisomy 13 is 2.3 in 10,000 births. Deletion cases have different types of genetic disorders, but the prognosis is much better than other chromosomal abnormalities. In European studies found 60% of cases of deletions and 90% of cases of microdeletions have live birth outcomes. Unlike the case with trisomy cases whose births are only 49% and polyploid cases which are much lower at only 3%.[17] In this study also obtained outcomes of live births on p13p15 deletion subjects with congenital abnormalities in the form of omphalocele.

It has been said that polyploid cases have a small live birth rate. Even if the baby is born alive, it will often die after birth until the age of 1 year. From a study found that the research subjects with tetraploids are the oldest 26 months old, so tetraploid cases are rare but very lethal cases.[29]

Congenital abnormalities from tetraploid cases are very diverse namely intrauterine hypotrophy, growth retardation, dysplastic ears, microcephaly and microphthalmia, facial abnormalities such as micrognathia, and abnormalities of the hands and feet. Congenital abnormalities in tetraploid cases are often unique and are rarely found in other types of chromosomal abnormalities, namely severe microphthalmia to anophthalmia, and meningomyelocele that are markers for clinicians in the diagnosis of tetraploid cases.[29] Likewise, this study found abnormalities in the extremities, although no tetraploid abnormalities were found that were typical on USG examination. The subject also has a termination outcome because the possibility of life is also small.

Table 6 shows we can see the similarity between amniocentesis, ultrasound, and birth abnormalities It is known from various studies that amniocentesis is a good examination to assess for congenital abnormalities. Based on research from Akbari et al, it was found that compared with biomarker screening from maternal serum, ultrasound translucency, chorionic villous sampling, and amniocentesis, it was concluded that amniocentesis had the highest diagnostic ability (> 99.9%) and with the lowest false positive (zero).[30] This can be seen from this study, where there were no patients with abnormalities on chromosome analysis and congenital anomalies at birth but there were no abnormalities on the USG examination.

Overall baby outcomes can be seen in Table 7, where 17 alive (53.1%), 13 died (40.6%), and 2 were terminated (6.3%). Of the 13 babies who died the majority had congenital abnormalities in the cardiovascular system (61.53%). Based on research from Gatt et al who examined the contribution of congenital abnormalities to neonatal mortality in Malta, congenital abnormalities accounted for 36.7% of neonatal deaths.[31] Whereas the research conducted by Roncancio et al who examined the characteristics of fetal and neonatal deaths due to congenital abnormalities in Colombia from 1999-2008 found that the most common congenital abnormalities resulting in death were cardiovascular abnormalities (32.0%), central nervous system abnormalities (15.8%) %) and chromosomal abnormalities (8.0%).[32]

The gender of 32 study subjects, obtained as many as 16 male babies (50%), 14 female babies (43.8%), and termination of 2 babies (6.2%) are shown in Table 9. Based on research from Ajao et al who examined the outcomes of infants with congenital abnormalities in Nigeria, 53.7% were male. Male gender is associated with a higher risk of death compared to the female gender. The comorbidities increase the risk of neonatal death but do not independently increase the risk.[33]

From this research and various reference studies, it can be concluded that the amniocentesis examination is a gold standard examination in assessing the presence of chromosomal abnormalities that cause various birth defects or defects. The best tests are done over the 15th week to increase the safety of the baby's output.

5. Conclusion

In conclusion, based on the results of descriptive research to determine the profile of pregnancies undergoing amniocentesis procedures in the SIDIC Fetal Maternal Outpatient Department in Sanglah Hospital from June 2017 to June 2019, there were 7 births were congenital abnormalities (0.31%) with chromosomal abnormalities out of a total of 2273 childbirth and 25 births of congenital abnormalities without chromosomal abnormalities (1.09%). The incidence of chromosomal abnormalities based on the analysis of 32 amniocentesis samples was 21,875% (7 samples). Seven chromosomal abnormalities profile obtained were trisomy 18, trisomy 13, tetraploid, and deletion. While the profile of abnormalities based on the most ultrasound examination is central nervous system abnormalities, cardiovascular system abnormalities, and gastrointestinal abnormalities. The baby outcome was 53.1% of live babies, 40.6% of died babies and 6.3% terminations

References

[1] Feldkamp ML, Carey JC, Byrne JLB, Krikov S, Botto LD. Etiology and clinical presentation of birth defects:

population based study. BMJ. 2017;357:j2249. Published 2017 May 30. doi:10.1136/bmj.j2249.

[2] American Colleague of Obstetrician and Gynecologits. Practice Bulletin No. 162. (2016). *Obstetrics & Gynecology, 127(5), e108–e122.* doi:10.1097/aog.0000000000001405.

[3] Blencowe H, Kancherla V, Moorthie S, Darlison MW, Modell B. Estimates of global and regional prevalence of neural tube defects for 2015: a systematic analysis. Annals of the New York Academy of Sciences. 2018 Jan 24.

[4] Kementerian Kesehatan RI. Infodatin: kelainan bawaan. Jakarta: Pusat Data Informasi Kementerian Kesehatan RI; 2018.

[5] St Louis AM, Kim K, Browne ML, Liu G, Liberman RF, Nembhard WN, Canfield MA, Copeland G, et al. Prevalence trends of selected major birth defects: A multi-state population-based retrospective study, United States, 1999 to 2007. Birth Defects Res. 2017 Nov 1;109(18):1442-1450. doi: 10.1002/bdr2.1113. Epub 2017 Sep 14.

[6] Egbe A, Uppu S, Lee S, Stroustrup A, Ho D, Srivastava S. Congenital malformations in the newborn population: a population study and analysis of the effect of sex and prematurity. Pediatr Neonatol. 2015;56:25–30.

[7] WHO South-East Asia (2013). *Birth defects in South-East Asia A public health challenge*. [online] Available at: http://apps.searo.who.int/PDS_DOCS/B4962.pdf [Accessed 25 Aug. 2019].

[8] Carlson, L. M., & Vora, N. L. (2017). Prenatal Diagnosis. Obstetrics and Gynecology Clinics of North America, 44(2), 245–256. doi:10.1016/j.ogc.2017.02.004 (https://doi.org/10.1016/j.ogc.2017.02.004).

[9] Who.int. (2019). *Congenital anomalies*. [online] Available at: https://www.who.int/news-room/fact-sheets/detail/congenital-anomalies [Accessed 31 Aug. 2019].Saldarriaga-Gil, 2014.

[10] Too, G. and Berkowitz, R.L. 2018. Cordocentesis and Fetal Transfusion. In: *Obstetric Imaging: Fetal Diagnosis and Care*. Elsevier, pp. 475-478.e1. Available at: https://linkinghub.elsevier.com/retrieve/pii/B9780323445481001121 [Accessed: 12 January 2020].

[11] WHO (2015). *Disease burden and mortality estimates: Child Causes Of Death, 2000–2016* [online] Available at:https://www.who.int/healthinfo/global_burden_disease/ estimates/en/index3.html [Diakses tanggal: 7 Desember 2019].

[12] Sallout B, Obedat N, Shakeel F, Mansoor A, Walker M, Al-Badr A. Prevalence of major congenital anomalies at King Fahad Medical City in Saudi Arabia: a tertiary care centre-based study. *Ann Saudi Med*. 2015;35(5):343–351. doi:10.5144/0256-4947.2015.

[13] Shanks, A., Odibo, A., Macones, G., Cahill, A., & Goetzinger, K. (2016). *Advanced Maternal Age and the Risk of Major Congenital Anomalies. American Journal of Perinatology, 34(03), 217–222.* doi:10.1055/s-0036-1585410.

[14] Zhang XH, Qiu LQ, Ye YH, Xu J. Chromosomal abnormalities: subgroup analysis by maternal age and perinatal features in zhejiang province of China, 2011-2015. *Ital J Pediatr*. 2017;43(1):47. Published 2017 May 12. doi:10.1186/s13052-017-0363-y.

[15] Alfirevic Z, Navaratnam K, Mujezinovic F. Amniocentesis and chorionic villus sampling for prenatal diagnosis. Cochrane Database of Systematic Reviews 2017, Issue 9. Art. No.: CD003252. DOI: 10.1002/14651858.CD003252.pub2.

[16] Wellesley D, Dolk H, Boyd PA, Greenlees R, Haeusler M, Nelen V, et al. Rare chromosome abnormalities, prevalence and prenatal diagnosis rates from population-based congenital anomaly registers in Europe. European Journal of Human Genetics. 2012; 20:521–526.

[17] Onyambu CK, Tharamba NM. Screening for congenital fetal anomalies in low risk pregnancy: the Kenyatta National Hospital experience. *BMC Pregnancy Childbirth*. 2018;18(1):180. Published 2018 May 23. doi:10.1186/s12884-018-1824-z.

[18] Eros FR, Beke A. Congenital Fetal Anomalies and the Role of Prenatal Ultrasound. 2017. Intech Open. DOI: 10.5772/intechopen.71907.

[19] Todros T, Capuzzo E, Gaglioti P. Prenatal diagnosis of congenital anomalies. *Images Paediatr Cardiol*. 2001;3(2):3–18.

[20] Sieroszewski P, Perenc M, Budecka E, Suzin J. Ultrasound diagnostic schema for the determination of increased risk for chromosomal fetal aneuploidies in the first half of pregnancy. Journal of Applied Genetics. 2006. 47(2):177-185.

[21] Wald, N.J. 2018. Prenatal reflex DNA screening for trisomy 21, 18 and 13. Expert Review of Molecular Diagnostics 18(5), pp. 399–401. doi: 10.1080/14737159.2018.1462703.

[22] Waltman, L. A., Eckel-Passow, J. E., Sharma, R. G., & Van Dyke, D. L. (2013). Advanced Maternal Age in Polyploidy With Concurrent Aneuploidy. American Journal of Medical Genetics Part A, 161(5), 1200–1202.

[23] Duong HT, Hoyt AT, Carmichael SL, et al. Is maternal parity an independent risk factor for birth defects?. Birth Defects Res A Clin Mol Teratol. 2012;94(4):230–236.

[24] Jawad S, Ikram Ul Haq, Cheema MR. Role of multiparity in birth defects. Professional Med J 2017;24(8):1241-1244.

[25] Savva, G.M.,Walker,K., &Morris,J.K. (2010).The maternal age-specificlivebirth prevalence of trisomies 13 and 18 compared to trisomy 21 (Downsyndrome). Prenatal Diagnosis,30,57–64.

[26] Wu, J., Springett, A., & Morris, J.K. (2013). Survival of trisomy 18 (Edwards syndrome) and trisomy 13 (Patau syndrome) in england and wales: 2004–2011. American Journal of Medical Genetics Part A,161A,2512–2518.

[27] Witters, G. et al. 2011. Trisomy 13, 18, 21, Triploidy and Turner syndrome: the 5T's. Look at the hands. Facts, Views & Vision in ObGyn 3(1), pp. 15–21.

[28] Bothur-Nowacka J, Jezela-Stanek A, Zaniuk K, Goryluk-Kozakiewicz B, Krajewska-Walasek M, Dobrzanska A. Tetraploidy in the era of molecular karyotyping – What we need to remember. Pediatria Polska. 2013;88(5):467-71.

[29] Akbari M, Mirmazloumi SH, Garshasbi M, Talari ZS, Sadeghi F. Specificity and Sensitivity of NIPT for Prenatal Screening of Down Syndrome in100 Pregnant Women from Tonekabon, Iran. iMedPub Jourals. 2018;4(1):1-3.

[30] Gatt M, England K, Grech V, Calleja N. Contribution of Congenital Anomalies to Neonatal Mortality Rates in Malta. *Paediatric and Perinatal Epidemiology*. 2015;29:401-406. Doi: 10.1111/ppe.12206.

[31] Roncancio CP, Misnaza SP, Pena IC, Prieto FE, Cannon MJ and Valencia D. Trends and Characteristics of fetal and neonatal mortality due to congenital anomalies, Colombia 1999-2008. *The Journal of Maternal-Fetal & Neonatal Medicine*. 2017. Published 2017 May 22. DOI : 10.1080/14767058.2017.1326901.

[32] Ajao AE, Adeoye IA. Prevalance, risk factors and outcome of congenital anomalies among neonatal admissions in OGBOMOSO, Nigeria. *BMC Pediatrics*. 2019;19(8):8. Published 2019 April 03. doi:10.1186/s12887-019-1471-1.