



## Characteristics of fetal congenital anomalies at outpatient clinic of tertiary hospital

Hermanto Tri Joewono <sup>1\*</sup>, Agus Sulistyono <sup>1</sup>, Ratriana Via Parasati <sup>2</sup>, Aditiawarman <sup>1</sup>

<sup>1</sup> Division of Maternal-Fetal Medicine, Department of Obstetrics and Gynecology, Faculty of Medicine-Dr. Soetomo Teaching Hospital, Universitas Airlangga, Surabaya 60131, INDONESIA

<sup>2</sup> Department of Obstetrics and Gynecology, Faculty of Medicine-Dr. Soetomo Teaching Hospital, Universitas Airlangga, Surabaya 60131, INDONESIA

\*Corresponding author: [hermanto.tri@fk.unair.ac.id](mailto:hermanto.tri@fk.unair.ac.id)

### Abstract

Congenital anomalies are a major cause of infant mortality and are an important contributor to child and adult morbidity. This study aimed to analyze the characteristics and outcomes of fetus with congenital malformation at tertiary hospital. This was a descriptive-retrospective study using secondary data from medical records, weekly reports, and perinatal death conference from January 2013 to December 2015. There were 82 congenital anomalies in 3 years with age of the mothers of 35 years old as many as 53 (60.9%), from outside Surabaya (n=54, 62.06%), and gestational age of 37-42 weeks in 43 women (52.44%). Non-booked congenital anomalies were found in 54 (65.8%) mothers. The types of anomalies found were biggest proportion hydrocephalus in 32 infants (36.78%), and multiple congenital anomalies in 17 infants (19.5%). The major sex of infants born was female as many as 40 (48.78%). Mode of deliveries was mostly by caesarean birth as many as 42 (51.73%). There were congenital anomalies with fetal congenital anomalies. The most common anomaly was hydrocephalus. The major sex of infants born was female and mode of deliveries was mostly by caesarean birth.

**Keywords:** congenital anomalies, fetal congenital malformation, infant mortality

Joewono HT, Sulistyono A, Parasati RV, Aditiawarman (2020) Characteristics of fetal congenital anomalies at outpatient clinic of tertiary hospital. *Eurasia J Biosci* 14: 1651-1655.

© 2020 Joewono et al.

This is an open-access article distributed under the terms of the Creative Commons Attribution License.

### INTRODUCTION

In Southeast Asia, from 320 per 100,000 live births in 1990 to 110 per 100,000 live births in 2012, the maternal mortality ratio (MMR) has decreased by 66 percent from 1990 to 2015. However, not all countries experienced this decline (Kalliora et al. 2018). The Indonesian MMR estimates for 2008 to 2012 were 359 per 100,000 live births (DeSilva et al. 2016). In 2015, 305 MMRs per 100,000 live births were reported (Nasution et al. 2019). Congenital anomalies in the growth of fetal structures arise from the life of the conception of the egg. Congenital anomaly is the most common cause of miscarriage and perinatal death. It is caused by various factors both genetic and non-genetic. For certain cases, women who have no family history of congenital anomalies and no identified risk factors give birth to these babies. Genetics, the environment, or some combination of the two are therefore thought to play a role in the development of congenital anomalies. Congenital abnormalities and related health care costs are also potentially modifiable, and environmentally contaminants released into the environment may be avoided. The relation between environmental contaminant exposure and the development of

congenital anomalies must therefore be better established (Foster et al. 2017).

In developing countries, maternal mortality is estimated at 100 to 1,000 or more per 100,000 live birth and 7 to 15 per 100,000 live birth in developed countries (Amiruddin et al. 2018). As from March 2014, congenital anomalies were published and comprehensively reviewed in more than 4000 pregnant women who received various types of vaccination against adjuvant and non-adjuvant influenza in the first trimester and over 19,000 in any trimester. The severity of congenital abnormalities varies greatly. Certain congenital anomalies in the early postnatal period are linked to spontaneous abortion, stillbirth, or death. World mortality due to congenital anomalies has decreased to 632,100 in 2013 from 750,600 in 1990, with age-standardized death rates of respectively 11.0 and 8.7 per 100,000. Congenital heart anomalies (323.4%), neural tube defects (68.9%), Down's syndrome (36.4%), and chromosomal unbalanced rearrangements (17.3%) are subtypes of fatal congenital anomalies (which were

Received: October 2019

Accepted: May 2020

Printed: June 2020

estimated in thousands in 2013). Other congenital anomalies may have little survival impact. Anomalies which affect the life expectancy of an infant, status of health, and physical or social function may be described as “major” anomalies. “Minor” anomalies, by contrast, have little or no health effect, or have a long or short duration of function. Because of their effect on the public health and current monitoring and reporting systems of major national and international organizations, we have chosen to concentrate on major anomalies in this congenital anomalies definition (DeSilva et al. 2016).

Across four centers, 20 maternal deaths occurred. In Hat Yai, Thailand (14.9/1000 births), the maternal death rate was the highest by the center, followed by Medan, Indonesia (14.1/1000), Hanoi, Vietnam (3.9/1000) and Padang, Indonesia (3.7/1000.) Eclampsia and other hypertensive conditions induced by pregnancy were most common, with 10 of those 20 deaths contributing. Major contributing causes include medical and congenital abnormalities of the mother (Chongsuvivatwong et al. 2010). Congenital anomaly is a major cause of infant mortality and is an important contributor to child and adult morbidity. The etiology of the majority of these congenital anomalies is still unknown. Prematurity, low birth weight, asphyxia, resuscitation during delivery, invasive procedure, congenital abnormalities, parenteral nutrition, long-term hospital hospitalization are risk factors of neonatal (Utomo 2010). Congenital abnormalities are having a significant impact in contributing to neonatal and infant mortality and morbidity, developed nations have formulated a detailed assessment method to discover the prevalence of congenital anomalies for the development of successful preventive measures. Congenital anomaly is a condition occurring at the moment of conception that differs from the normal presentation. In 2004, WHO estimated that about 260,000 deaths worldwide were triggered by congenital anomalies or defects (Kalliora et al. 2018; Ivanov et al., 2019). This is nearly seven percent of all neonatal deaths. It is projected that 94 percent of severe birth defects exist in middle and low-income countries where mothers are more vulnerable to macro-and micronutrient deficiency and have greater vulnerability to perinatal infection (Ahmed et al. 2017).

Fetal anomaly screening is the most successful way of minimizing the incidence of severe congenital abnormalities and growing the recovery rate of children born with these conditions. The detection of a correctable abnormality can mean that delivery may take place in a pediatric surgery facility, and the identification of a serious uncorrectable abnormality that result in the offering of termination of pregnancy. Survivors with congenital anomalies may have life-long physical, behavioral, sensory and auditory disorders if not adequately handled, and can adversely impact the personal and economic existence of the individual

**Table 1.** Congenital Anomalies Patients Characteristics

Characteristic	Total	Percentage
<b>Domicile</b>		
Surabaya	28	37.94%
Outside Surabaya	54	62.06%
<b>Age</b>		
< 17	1	1.21%
17-35	2	34.14%
>35	53	60.91%
<b>Gestational Age</b>		
<37 weeks	39	47.56%
37-42 weeks	43	52.44%
>42 weeks	0	0
<b>Number of Antenatal Patient's Treatment</b>		
<3 times	54	65.8%
>3 times	28	34.2%

affected, as well as their families and societies (Ameen et al. 2018). Many causes of maternal death can be prevented or managed with evidence-based healthcare services that are available in Indonesia. This study aimed to find out more about the prevalence, socio-demographic characteristics, and characteristics of infants with congenital anomalies in the Outpatient Installation of Dr. Soetomo Hospital, Surabaya.

## MATERIAL AND METHODS

This was a descriptive study of infant congenital anomaly in Dr. Soetomo Hospital, Surabaya from January 1, 2013 to December 31, 2015. Data were taken from medical records of outpatients in Dr. Soetomo Hospital, Surabaya. The diagnosis of infant congenital anomaly was determined based on an ultrasound examination in the fetomaternal field of Dr. Soetomo Hospital, Surabaya. There were 137 congenital anomalies of infants with congenital anomalies found from January 1 2013 to December 31, 2015. Five congenital anomalies were not include because they were born outside Dr. Soetomo Hospital, Surabaya. Data was analyzed descriptively.

## RESULTS

There were 137 congenital anomalies of infants with congenital anomalies found from January 1 2013 to December 31, 2015. Five congenital anomalies were not include because they were born outside Dr. Soetomo Hospital, Surabaya. **Table 1** shows that 82 congenital anomalies include in the study. A total of 53 congenital anomalies was found in patients aged over 35 years (60.91%).

**Table 2** shows that the majority of patients (n=54, 65.8%) visit less than 3 times at the Obstetric Polyclinic of Dr. Soetomo. The most congenital anomaly was hydrocephalus as many as 32 (36.78%), and multiple congenital anomalies were 17 (19.5%) (**Table 5**). The majority of infants' sex was female as many as 40 (48.78%).

**Table 2.** Type of Congenital Anomalies

Characteristic	Total	Percentage
<b>Type of Congenital Anomalies</b>		
Hydrocephalus	32	39.02%
Holoprosencephaly	4	4.87%
Fetal Hydrops	10	12.19%
Anencephaly	6	7.31%
Omphalocele	5	6.09%
Meningocele	4	4.87%
Diaphragmatic Hernia	4	4.87%
Multiple	17	20.73%
<b>Gender</b>		
Male	36	43.9%
Female	40	48.78%
Unknown	6	7.31%
<b>Delivery Mode</b>		
Vaginal Delivery	40	48.27%
Caesarean Section (CS)	42	51.73%

## DISCUSSION

Most of the Bartholin gland disorders patients who came to the obstetric center of Dr. Soetomo Hospital came from Surabaya. This may be due to patients from out of town who had been treated by a local obstetrician and only congenital anomalies with severe complications were referred to Surabaya. From the data we found, the congenital anomaly was found in women aged >35 years. Patients over the age of 35 do have a risk of discordance and infants due to chromosomal congenital anomaly. The incidence of post-term labor is influenced by some factors. Research conducted indicates that the risk of labor events post term or childbirth at gestational age  $\geq 42$  weeks are higher in women with age (>35 years old), have a heavy body excess, primipara, or have a history previous post term deliveries. The major sex of infants born was female and mode of deliveries was mostly by caesarean birth. Cesarean section delivery is intended to reduce maternal and fetal mortality and with high birth rates and the limited obstetric health resources of Asian developing countries (Lumbaraja 2013). Other studies conducted well mentioned that maternal age is risky 45.20% had a post term pregnancy. This is related to the incomplete maturity of the reproductive organs in mothers aged <20 years and decreased function of the maternal organs >35 years (Maulinda Nadhifa Anwar 2018).

Information of common biological processes for congenital abnormalities and cancer is minimal, although the suggested causes involve non-genetic factors (e.g., environmental factors) that contribute to both disorders, somatic mutations in early embryogenesis genes that contribute to tissue mosaicism or chromosome microdeletions that comprise both developmental and cancer predisposition genes. The biological underpinnings of these associations are likely to differ by particular birth anomalies and different cancer types. Children with certain forms of congenital defects can die prematurely and thus lack the ability to

develop childhood cancer, which may attenuate our association (Norwood et al. 2017).

In this research, a significant correlation was observed between a family history of congenital anomalies and the occurrence of new congenital anomalies. Our finding is consistent with the results of a previous study that in subsequent pregnancies, the presence of a congenital disorder linked to a chromosomal abnormality, whether live-born or stillborn, raised the risk of chromosomal defects (Ameen et al. 2018). Chromosomal microarray analysis (CMA) has been widely used in the clinical assessment of patients with developmental delay, intellectual disability, multiple congenital anomalies, and autism spectrum disorders. Various studies have indicated that more widespread fetal CMA monitoring would lead to greater detection of clinically significant chromosome abnormalities. The American College of Obstetricians and Gynecologists and the Society for Maternal-Fetal Medicine note that prenatal CMA is most useful when ultrasonography assessments detect fetal structural abnormalities (Peng et al. 2016). Maternal mortality rate is the number of mothers' mortality that occurs during pregnancy, labor and childbirth. Maternal mortality is a big concern because it leads the state to lack other competitive resources, increasing infant morbidity and mortality (Fitriani et al. 2019).

This study showed that data from Dr. Soetomo Hospital was mostly born in pregnancy with most of the anomaly in the form of hydrocephalus with a good prognosis and with gestational age at term, so it was decided to be born in vaginal birth. Congenital anomalies are generally known as single-system or multiple-system malformations. The first type involves a particular organ system or body part, and the second type involves more than one organ system or body component. Major congenital defects are described as those that, if not corrected, may result in a severe loss in normal body function or even a decrease in life expectancy. Minor congenital anomalies involve defects that do not cause disability or have major physical or functional consequences and may be assumed to be normal variations (Ameen et al. 2018).

One was that the research aimed to diagnose more external (overt) congenital abnormalities in neonates during the first 48 h of birth, depending solely on clinical evaluations to establish a diagnosis. Neither cytogenetic analysis nor stillbirth autopsies have been conducted as such treatments are costly and have minimal supply in our locality. It is also probable that the research skipped certain congenital anomalies that did not arise early in life, such as cardiac disorders, pyloric stenosis, and urinary system anomalies, which may also justify the small number of deficiencies observed in contrast to other studies (Ameen et al. 2018).

There is much literature about the impact of infant loss on parents. However, most of it focuses on death

through sudden and unexpected means such as Sudden Infant Death Syndrome (SIDS), stillbirth, extreme prematurity, or miscarriage. The experience of parents whose infant has a life-limiting congenital anomaly is different. These parents know their babies will die. Scant research exists in which authors explored the experience of parents who had an infant die from life-limiting congenital anomalies. Another study showed babies lived 48 hours to 15 months of age. Many of these babies only lived a few hours after birth. Thus, it would be important to explore the experience of parents whose babies lived less than 48 hours to see if that experience mirrored those whose babies lived longer (Bush Welch 2018).

The frequency of sex differences in congenital anomalies is not significantly different. However, the data that we obtained in 40 congenital anomalies were found in the female sex at Dr. Soetomo Hospital, Surabaya. Sex distinctions in the incidence of congenital abnormalities have been identified for decades. In 1971, reported research on 14 congenital anomalies established that males had a higher prevalence of 8/14 congenital anomalies, and females had a higher prevalence of 4/14 congenital anomalies, with just 2 congenital anomalies showing a comparable prevalence between sexes. Numerous research have subsequently validated this finding and all conclude that the majority of congenital defects are more common in males than females, but these population-based findings draw results from small sampling ranges and are not contemporary (Black et al. 2020).

Theoretically, congenital defects resulting from an organ that grow equally in both males and females will arise at a comparable pace in both sexes. However, there has been a long history of discrepancy between the prevalence of congenital anomalies in males and females. Whereas, the average incidence of congenital abnormalities has been recorded to be 2.8% in females, this figure is 40% higher (overall incidence = 3.9%) in

males. Another research also found that males had a 29 percent greater chance of congenital defects relative to their twin sisters. Several subsequent population-based studies have identified this trend and confirmed the finding that some congenital anomalies are more common in males, while others are more common in females. However, congenital anomalies usually have a low event rate; therefore, studies based on population often have relatively small sample sizes, most of which have less than 1000 congenital anomalies identified (Black et al. 2020).

Alternatively, the difference in embryonic growth between the sexes is more likely to be clarified. Several researches analyzed the human sex ratio from conception to birth and noticed that the overall female mortality during pregnancy exceeds the overall male mortality. This suggests that a greater proportion of malformed females do not survive after conception, leading to a higher prevalence of malformed males in live births. Such results are compatible with those that indicate that males are more susceptible to organogenesis abnormalities than females, who are more vulnerable during the blastocyst period. Vulnerability during the blastocyst period results in pregnancy complications, although vulnerability during organogenesis results in a higher occurrence of congenital anomaly survival (Orzack et al. 2015).

## CONCLUSION

This study obtained congenital anomalies of infants in Dr. Soetomo General Hospital with maternal age when the mothers were pregnant over 35 years. Gestational age when mothers arrived was 37-42 weeks. Patients constituted 54 non-booked congenital anomalies. The major congenital disorder was hydrocephalus, and the most infants born were female with congenital anomalies which were delivered through abdominal.

## REFERENCES

- Ahmed W, Dey D, and Farid R, (2017) Prevalence and Pattern of Congenital Anomalies and Its Outcome at Chattagram Maa-O-Shishu General Hospital. *Chattagram Maa-O-Shishu Hospital Medical College Journal* 16(1): 22–25. <https://doi.org/10.3329/cmshmcj.v16i1.34981>
- Ameen SK, Alalaf SK, and Shabila NP, (2018) Pattern of congenital anomalies at birth and their correlations with maternal characteristics in the maternity teaching hospital, Erbil city, Iraq. *BMC Pregnancy and Childbirth* 18(1): 1–8. <https://doi.org/10.1186/s12884-018-2141-2>
- Amiruddin H, Ansariadi, and Palutri S, (2018) Counseling quality of danger signs pregnancy in work region or rural and urban primary health center district Jeneponto. *ACM International Conference Proceeding Series* 99–104. <https://doi.org/10.1145/3242789.3242818>
- Black AJ, Lu DY, Yefet LS, and Baird R, (2020) Sex differences in surgically correctable congenital anomalies: A systematic review. *Journal of Pediatric Surgery* 10. <https://doi.org/10.1016/j.jpedsurg.2020.01.016>
- Bush Welch S, (2018) Navigating Infant Death from Life-Limiting Congenital Anomaly: A Classic Grounded Theory Study. *The Grounded Theory Review* 17(1): 7.



- Chongsuvivatwong V, Bachtiar H, Chowdhury ME, Fernando S, Suwanrath C, Kor-Anantakul O, et al., (2010) Maternal and fetal mortality and complications associated with cesarean section deliveries in teaching hospitals in Asia. *Journal of Obstetrics and Gynaecology Research* 36(1): 45–51. <https://doi.org/10.1111/j.1447-0756.2009.01100.x>
- DeSilva M, Munoz FM, Mcmillan M, Kawai AT, Marshall H, Macartney KK, Joshi J, Oneko M, Rose AE, Dolk H, Trotta F, Spiegel H, Tomczyk S, Shrestha A, Kochhar S, and Kharbanda EO, (2016) Congenital anomalies: Case definition and guidelines for data collection, analysis, and presentation of immunization safety data. *Vaccine* 34(49): 6015–6026. <https://doi.org/10.1016/j.vaccine.2016.03.047>
- Fitriani, Masni, Bustan MN, Salmah U, Stang, and Syafar M, (2019) Analysis of risk factors maternal mortality in Sinjai District. *Indian Journal of Public Health Research and Development* 10(10): 1503–1507. <https://doi.org/10.5958/0976-5506.2019.03050.X>
- Foster WG, Evans JA, Little J, Arbour L, Moore A, Sauve R, Andrés León J, and Luo W, (2017) Human exposure to environmental contaminants and congenital anomalies: a critical review. *Critical Reviews in Toxicology* 47(1): 59–84. <https://doi.org/10.1080/10408444.2016.1211090>
- Ivanov DO, Iurev VK, Shevtsova KG, Moiseeva KE, Berezkina EN (2019) Fetoinfantile losses in the north-west region of Russia. *Electronic Journal of General Medicine*, 16(2): em135. <https://doi.org/10.29333/ejgm/102417>
- Kalliora C, Mamoulakis C, Vasilopoulos E, Stamatiades GA, Kalafati L, Barouni R, Karakousi T, Abdollahi M, and Tsatsakis A, (2018) Association of pesticide exposure with human congenital abnormalities. *Toxicology and Applied Pharmacology* 346(December 2017): 58–75. <https://doi.org/10.1016/j.taap.2018.03.025>
- Lumbanraja SN, (2013) Determining the maternal characteristics that predicts the adverse outcomes for patients with preeclampsia. *Journal of the University of Malaya Medical Centre* 16(1): 1–6.
- Maulinda Nadhifa Anwar RT, (2018) The Relationship between Age and Maternity Parity with Postterm Birth. The Influence of Patients' Characteristics with Intravena Catheter in Phlebitis Incidence Nella 6: 1–8. <https://doi.org/10.20473/jbe.v6i12018.27-34>
- Nasution SK, Mahendradhata Y, and Trisnantoro L, (2019) Can a National Health Insurance Policy Increase Equity in the Utilization of Skilled Birth Attendants in Indonesia? A Secondary Analysis of the 2012 to 2016 National Socio-Economic Survey of Indonesia. *Asia-Pacific Journal of Public Health*. <https://doi.org/10.1177/1010539519892394>
- Norwood MS, Lupo PJ, Chow EJ, Scheurer ME, Plon SE, Danysh HE, Spector LG, Carozza SE, Doody DR, and Mueller BA, (2017) Childhood cancer risk in those with chromosomal and non-chromosomal congenital anomalies in Washington State: 1984-2013. *PLoS ONE* 12(6): 1–15. <https://doi.org/10.1371/journal.pone.0179006>
- Orzack SH, Stubblefield JW, Akmaev VR, Colls P, Munné S, Scholl T, Steinsaltz D, and Zuckerman JE, (2015) The human sex ratio from conception to birth. *Proceedings of the National Academy of Sciences of the United States of America* 112(16): E2102–E2111. <https://doi.org/10.1073/pnas.1416546112>
- Peng R, Zhou Y, Xie HN, Zheng J, Xie YJ, and Yang JB, (2016) MCDA twins with discordant malformations: submicroscopic chromosomal anomalies detected by chromosomal microarray analysis and clinical outcomes. *Prenatal Diagnosis* 36(8): 766–774. <https://doi.org/10.1002/pd.4859>
- Utomo MT, (2010) Neonatal Sepsis in Low Birth Weight Infants in Dr. Soetomo General Hospital. *Indonesian Journal of Tropical and Infectious Disease* 1(2): 86. <https://doi.org/10.20473/ijtid.v1i2.2172>