Research Article

Demographic Characteristics of Mothers who Delivered Children with Birth Defects

Demografi Ibu yang Melahirkan Janin dengan Kelainan Bawaan

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Abstract

Objective: To determine the characteristics and background of mothers who delivered neonates with birth defects.

Methods: A retrospective study was used by evaluating the medical records of patients with birth defects in Dr. Cipto Mangunkusumo Hospital during the period between September 2014 to June 2016.

Results: A total of 67 (1.85%) out of 3,619 infants who were born in Dr. Cipto Mangunkusumo Hospital during the period between September 2014 and June 2016 had birth defects. Forty-seven (70.1%) mothers of the subjects irregularly attend antenatal care. The most frequent maternal comorbid disease in this study was asthma, which was found in 4 (5.97%) mothers of the subjects. 48 (58.7%) subjects had birth weight under 2500 g.

Conclusion: In this retrospective study, the main highlight is that 70.1% of the mothers who delivered neonates with birth defects did not attend antenatal care regularly. 58.7% of the neonates with birth defects had low birth weight. This study could be used as a base for further research investigating the role of antenatal care in early detection and/or the planning of delivery for babies with birth defects. Trends in babies with birth defects suggested that fetuses diagnosed with IUGR/SGA should be given special attention, as they were at increased risk for birth defects.

[Indones J Obstet Gynecol 2018; 6-4: 222-227]

Keywords: birth defect, maternal characteristics

Abstrak

***Tujuan**: Untuk mengetahui deskripsi/ciri-ciri dan latar belakang ibu yang melahirkan janin dengan kelainan bawaan.*

***Metode**: Studi retrospektif digunakan dengan menggunakan data sekunder rekam medis pasien dengan kelainan bawaan di Rumah Sakit Umum Pusat Nasional Dr. Cipto Mangunkusumo pada periode September 2014-Juni 2016.*

***Hasil**: Sejumlah 67 (1,85%) dari 3.619 neonatus didapatkan dengan kelainan bawaan di RSUPN Dr. Cipto Mangunkusumo pada periode September 2014-Juni 2016 . Sejumlah 47 (70,1%) ibu dari subjek tidak teratur dalam melakukan kunjungan antenatal care. Penyakit komorbid ibu yang paling banyak ditemukan dalam studi ini adalah asma, yang ditemukan dalam 4 (5,97%) subjek. 48 (58,7%) subjek memiliki berat lahir di bawah 2500 g.*

***Kesimpulan**: Pada studi retrospektif ini didapatkan 70,1% ibu yang melahirkan bayi dengan kelainan bawaan tidak melakukan kunjungan antenatal care. Didapatkan bahwa 58,7% bayi yang lahir dengan kelainan bawaan memiliki berat badan lahir rendah (BBLR). Studi ini dapat dijadikan sebagai landasan dilakukannya studi yang lebih besar untuk mengevaluasi peran antenatal care terhadap deteksi dini dan/atau perencanaan persalinan bayi dengan kelainan bawaan. Tren pada bayi dengan kelainan bawaan menunjukkan bahwa janin yang pada antenatal care didapatkan IUGR/SGA patut diperhatikan lebih untuk kecurigaan kemungkinan adanya kelainan bawaan.*

[Maj Obstet Ginekol Indones 2018; 6-4: 222-227]

***Kata kunci**: deskripsi ibu, kelainan bawaan*

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INTRODUCTION

Embryonic development is a complex process from the time of fertilisation to the formation of cells, tissues and organs. In early pregnancy, each body organ (system) has a critical period of organogenesis. Interference during this early pregnancy with intrinsic and extrinsic factors (i.e. parental and multifactorial effects) may lead to different types of birth defects.[1-3] Birth defects are one of the major causes of infant and child mortality, morbidity, and long-term disability.[4,5] The term 'birth defects; refer to any morphological, functional, behavioural, and metabolic defects that develop during the organogenesis period and present at birth or detected later in life.[2] Birth defects may be caused by genetic, chromosomal, environmental, and multifactorial effects, as well as micronutrient deficiencies or unknown etiological agents.[6,7]

Birth defects affect 3% of all neonates, cause 3.2 million birth defect-related disabilities, and result in 2.7 million infant deaths.[8] They have a significant effect on individuals, families, health-care systems and societies. Birth defects are particularly serious health burden in low- and middle-income countries, including Indonesia, due to consanguineous marriage, advanced maternal age, migration and poverty.

The magnitude of birth defects varies from country to country and from race/ethnicity, and about 40-60% of their causes are unknown.[9] According to the World Health Organization, approximately 3 million fetuses and infants are born each year with major malformations.[2]

Approximately 94% of infants born with birth defect were reported to come from the middle and low-income countries, and it also recorded 95% of the death of such children from birth defects.[10] The defects pose serious psychological stress or nursing mothers due to potential life-long disability.[11]

Studies describing maternal characteristics associated with birth defects have never been conducted in Indonesia. We aim to describe the characteristics of mothers who give birth to infants with birth defects.

METHODS

A retrospective study design was used by evaluating the medical records of patients with birth defects in Dr. Cipto Mangunkusumo Hospital during the period between September 2014 and June 2016. Inclusion criteria include subjects with spina bifida, anencephaly, meningo/encephalocele, congenital cataract, cleft palate, cleft lip, cleft lip and palate, hypospadias, epispadias, talipes, reduction deformity, atresia ani with/without fistula, omphalocele, gastroschisis, or conjoined twins.

RESULTS

Sixty-seven (1.85%) out of 3,619 neonates born at Dr. Cipto Mangunkusumo Hospital during the period between September 2014 and June 2016 had birth defects. Six (9%) mothers of the subjects were Javanese. Fifteen (22.39%) mothers of the subjects resided in East Jakarta. Forty-seven (70.1%) mothers of the subjects irregularly attend antenatal care. Demographic characteristics of the subjects are presented in Table 1.

Table 1. Demographic Characteristics of the Mothers of the Subject

Characteristic	n (total 67)	%
Maternal ethnic group		
Javanese	6	9
Sundanese	3	4.5
Betawi	1	1.5
Batak	1	1.5
Padang	1	1.5
Badui	1	1.5
Others	1	1.5
Unknown	53	79.1
Domicile		
Central Jakarta	5	7.46
East Jakarta	15	22.39
West Jakarta	6	8.96
North Jakarta	3	4.48
South Jakarta	5	7.46
Bekasi	12	17.91
Tangerang	4	5.97
Depok	5	7.46
Bogor	1	1.49
Other	1	1.49
Unknown	10	14.93
Education		
Undergraduate	2	3
D3	1	16.4
Senior High School	11	1.5
Junior High School	1	1.5
Elementary	1	1.5
Unknown	51	76.1
Regularity of Antenatal Care Visits		
Regular	8	12
Irregular	47	70.1
Unknow	12	17.9
Antenatal Care Location		
Dr. Cipto Mangunkusumo Hospital	6	9
Other than Dr. Cipto Mangunkusumo Hospital	49	73.1
Maternal Age		
≤ 35 y.o	49	73
> 35 y.o	18	27
Gravidity		
< 2	22	32.84

≥ 2	45	67.16
Types of delivery		
Caesarean section	42	63
Spontaneous	25	37
Gestational age		
< 37 weeks	40	60
≥ 37 weeks	27	40
Maternal comorbid disease		
Asthma	4	5.97
Diabetes	1	1.49
Graves'disease	2	2.99
Others*	4	5.97
Denied	43	64.18
Unknown	13	19.40

**Others = severe preeclampsia, primary infertility, thyroid carcinoma, uterine myoma*

In this study, the commonest birth defect was omphalocele (20.48%), followed by labiognatopalatochizis (9.64%), talipes (9.64%), and others consisting of anencephaly, atresia ani, as well as conjoined twins (8.43%). Defects related to gastrointestinal system (29.79%) were the commonest, followed by central nervous system (19.7%), and facial and oral malformations (13.64%). The occurrence of multiple organ defects was 18.18%. Forty-two (63%) mothers of the subjects underwent caesarean section. Forty (60%) mothers had gestational ages below 37 weeks. Four (5.97%) mothers of the subjects had asthma in this study.

The percentage of each birth defect based on organ system were respectively as follow. The percentage of defects related to central nervous system including anencephaly, encephalocele, meningoencephalocele, ventriculomegaly, and spina bifida was 38.89%, 5.56%, 16.67%, 5.56%, and 33.33%, respectively. The percentages of defects related to gastrointestinal system including omphalocele, gastroschisis, and atresia ani, were 60.71%, 14.29%, and 25%, respectively. Musculoskeletal defects including talipes, limb shortness, hypoplasia of the extremities, and amelia were 57.14%, 28.57%, 7.14%, and 7.14%, respectively. Genitourinary defect including hypospadias was 5.97%. The percentages of other birth defects consisting of congenital cataract and conjoined twins were 12.5% and 87.5%.

Table 2. The Distribution of Prenatal Ultrasound and Postpartum Findings

Conformity	n
Prenatal ultrasound findings matching postpartum findings	36
Prenatal ultrasound findings not matching postpartum findings	12
No prenatal ultrasound findings	19

Of 67 subjects, 19 were excluded due to the lack of ultrasound findings. Of 48 subjects, 36 (75%) of prenatal ultrasound findings were matching with postpartum findings.

Table 3. Demographic Characteristics of the Subjects

Characteristic	n	%
Sex (n=67)		
Male	32	47.7
Female	35	52.3
Birth weight/ g (n=46)		
2500	19	41.3
<2500	48	58.7

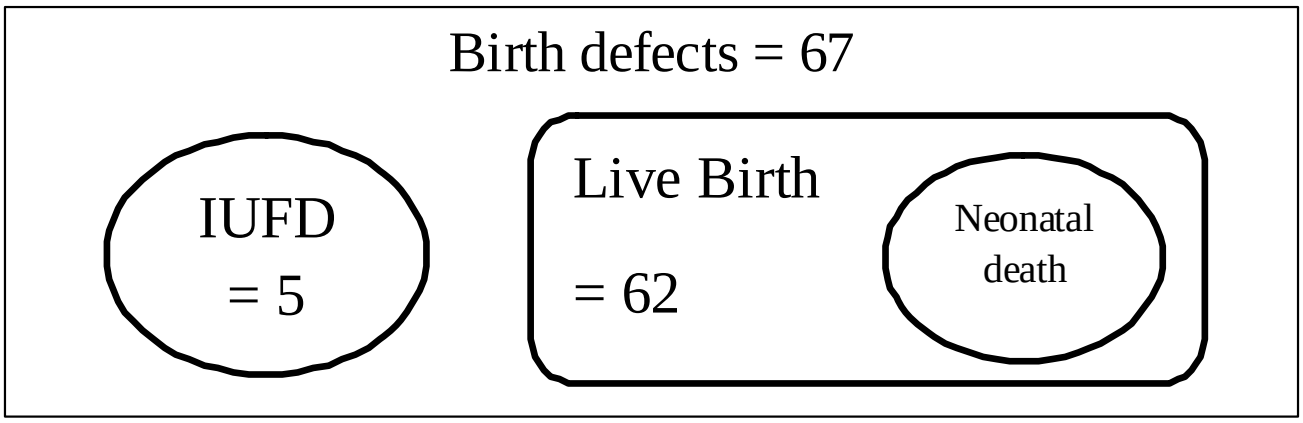

Figure1. The incidence of intrauterine fetal death (IUFD) and neonatal death.

The majority of the newborns were female (52.3%). 58.7% of the subjects had low birth weight (<2500 g). The mean birth weight of newborns was 2236.98 ± 792.92. The lowest and highest birth weights were 600 g and 3880 g, respectively. The incidence of intrauterine fetal death (IUFD) and neonatal death in their first 28 days of life were 7.46% and 32.8%, respectively (Figure 1).

DISCUSSION

Regularity of Antenatal Care Visits

Antenatal care is a crucial element for improving maternal and neonates health. Effective antenatal care improves maternal health through early detection, prevention as well as treatment of medical and obstetrical complications during pregnancy.[11] In this study, 40 (70.1%) mothers of the subjects irregularly attend antenatal care, which suggested that the regularity of antenatal care visits might be a key element in preventing birth defects. However, to our knowledge, studies investigating the association between regularity of antenatal care visits and the occurrence of birth defects have never been conducted. A possible explanation is that mothers who irregularly attend antenatal care have lower knowledge about the prevention of birth defects, which may lead to high incidence of birth defects. Further studies are required to investigate this association.

Maternal Educational Level

In this study, no direct association between maternal educational level and the occurrence of birth defects were found. This is in line with a previous study conducted by Bello et al[12] which revealed that the level of education had no significant relationship with their specific knowledge, knowledge in relation to risk factors and the overall knowledge about birth defect.

Maternal Age

In this study, maternal age above 35 years old had no direct association with the occurrence of birth defects. This is contrary to previous studies, which might be due to smaller population size. Hollier et al[13] concluded that increased maternal age was significantly associated with chromosomal aberrations, particularly aneuploidies. In addition, it has been reported that woman who were 25 years of age or older at delivery had significantly and progressively greater risk of having fetuses with non-chromosomal malformation compared to women aged 20-24 years. The National Center for Health Statistics[14] conducted a study about the effects of advanced maternal age on risks of congenital malformations. These data demonstrated a significant trend of increasing congenital heart disease with advancing maternal age. However, infants with chromosomal abnormalities were not considered separately in that study.

Gestational Age

In this study, 60% of the subjects had gestational age less than 37 weeks. This showed that the occurrence of birth defects is higher in the premature population. This finding is in line with several previous studies. In a population-based cohort study of 264,392 infants, 7,738 (2.93%) were identified as having birth defects. The study revealed that premature infants were more than two times as likely to develop birth defects (risk ratio [RR] = 2.43; 95% CI 2.30-2.56).[15]

Maternal Comorbid Disease

The most frequent maternal comorbid disease in this study was asthma (5.97%). This is in line with a previous study conducted by Blais et al[16], which found that maternal asthma was significantly associated with an increased risk of congenital malformation. This might be due to impaired fetal oxygenation and medications used to treat asthma. A systematic review and meta-analysis of 21 studies[17] found that maternal asthma was associated with a significantly increased risk of congenital malformations (RR 1.11, 95% CI 1.102-1.21).

Prenatal Ultrasound and Postpartum Findings

Prenatal ultrasound was established in Russia in 2000 as a routine method of screening for birth defects. The effectiveness of prenatal screening can be estimated by the prenatal detection rate, which represents the proportion of birth defects recognised before delivery.[20] The sensitivity and specificity of ultrasound were 39% and 99.9%, respectively. Several birth defects can be identified accurately based solely on prenatal findings. Exam-

ples include anencephaly, spina bifida, chromosomal abnormalities, and conjoined twins. However, not all birth defects can be identified accurately based solely on prenatal findings. The positive predictive value of prenatal ultrasound for congenital heart defects varies from 70% to 98%, depending on the type of ultrasound (four-chamber view alone, with outflow tract view, fetal echocardiography) and the specific cardiac defect.[18-21] In this study, 25% of the subjects' abnormalities were not identified accurately.

Birth Weight

In this study, the majority of the subjects had birth weight <2500 g. This finding is in line with previous studies. In a cohort of 307 fetuses with congenital heart diseases, 17 % were associated with a birth weight centile <10th when standard population centiles were used. The Baltimore-Washington Infant Study[22] suggested that, in infants with CHD, birth weight was significantly lower than the control population. Mili et al[23] found that low-birth-weight infants were at a 1.76-times higher risk of having birth defects than those weighing 2500 g to 3999 g.

Postnatal Treatment

Neonatal screening for birth defects may facilitate early detection, treatment and care. Neonatal screening programmes (physical examination of all neonates and screening for congenital hypothyroidism, phenylketonuria, sickle-cell disease and glucose-6-phosphate dehydrogenase deficiency) and training of primary health-care providers aids the diagnosis and appropriate referral for treatment of infants with birth defects. Physical examination of all neonates by trained primary health-care practitioners is feasible in most health care centre and allows the identification of numerous birth defects, including cardiovascular defects that are associated with a high risk of early mortality and referral.[8]

CONCLUSION

In this retrospective study, the main highlight is that 70.1% of the mothers who delivered neonates with birth defects did not attend antenatal care regularly. 58.7% of the neonates with birth defects had low birth weight. This study could be used as base for further research investigating about the role of antenatal care in early detection and/or the planning of delivery for babies with birth defects. Trends in babies with birth defects suggested that fetuses diagnosed with IUGR/SGA should be given special attention, as they were at increased risk for birth defects.

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