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ETHICS AND LEGAL ASPECT OF TERMINATION OF PREGNANCY WITH TRISOMY 13 (PATAU SYNDROME)

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ABSTRACT

Trisomy 13 is a serious genetic anomaly in the fetus that is one of the causes of abortion as a result of the chromosomal aneuploidy. Trisomy 13 affects roughly 1 in 10,000 to 20,000 live births, and more than 95% of pregnancies end in prenatal death. Due to the higher prevalence of preeclampsia and the danger of maternal death associated with early birth, abnormalities in these infants also have an impact on the mother's health. There is a medical emergency condition to save the mother's life in trisomy 13 pregnancy. Termination of pregnancy avoids the adverse effects that will be experienced by babies with severe congenital abnormalities. According to Law No. 36 of 2009 concerning Health, women who eligble for abortions if there are medical reasons to do so. Pregnancies that endanger the fetus' life and health, including those with severe genetic diseases that cannot be treated so that they have a negative impact on the baby and mother's life are medically indicated for medical provocatus abortions. The problems in this paper are 1) What are the ethical problems found in pregnancies with a fetus diagnosed with a genetic disorder due to trisomy 13? 2) What is the procedure for implementing medical abortion provocation that fulfills ethical aspects and complies with applicable regulations. To answer these problems, a research using normative legal research, accompanied by reports of cases of abortion. This normative legal research focuses on ethical and legal aspects of the implementation of provocative medical abortion following statutory regulations. This normative legal research starts with articles and case reports of fetal pregnancies with genetic disorders from the Gatot Soebroto Army Hospital. The results of the study show that consent to termination of pregnancy with medical indications by the patient and approved by the

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husband/family. Termination of pregnancy in a fetus with a severe genetic disorder Trisomy 13 is a legal abortion if it is carried out according to medical indications and steps regulated in Health Law No. 36 of 2009 and Government Regulation No. 61 of 2014 concerning Reproductive Health.

KEYWORDS *Trisomy 13, Medical Provocatus Abortion, informed consent, legal certainty, legal protection.*

INTRODUCTION

In Indonesia, there is a lot of discussion around the issue of abortion. According to data from the Indonesian Ministry of Health, thousands of unintended pregnancies occur each year, whether as a result of rape, wrongful affiliations that result in unmarried pregnancies, or failure of contraception.

Because most abortions are performed by couples who conceive outside of marriage, abortion is typically seen as a shame that needs to be kept hidden. Abortion is frequently prompted by unwanted pregnancies. Health professionals should pay extra attention to abortion since it is a risky medical procedure and involves moral quandaries. According to Bearak (2020) in 2015–19, there were 121.0 million unwanted pregnancies each year. There are 64 unwanted pregnancies per 1000 women aged 15–49 years. 61% of unintended pregnancies end in abortion (a total of 73·3 million abortions each year) (Bearak et al., 2020). From this number, it can also be calculated that three out of 10 pregnancies end in abortion.

According to 2014 research that looked at statistics on maternal mortality from the years 2003 to 2012, unsafe abortions were responsible for 7.9% of maternal fatalities, or the equivalent of 193,000 pregnancies, with an estimated value range of 4.7% - 13.2% Maternal Mortality Rate (MMR). Yet, data on the causes of MMR related to unsafe abortion can also be considered to not accurately reflect the reality because issues go unreported, specifically because abortion is still illegal under current legal laws.

Although though abortion is legal in some situations under state law, reporting an abortion is nonetheless difficult due to cultural and religious stigmas. Bleeding, infection, sepsis, genital damage, and necrotic bowel are the main factors that contribute to unsafe abortion fatalities (Haddad & Nour, 2009).

In Indonesia, few research have been able to document safe abortion practices; this is because of legal restrictions that still categorically forbid all activity linked to abortion. Only two studies have attempted to demonstrate the estimated 1000 abortions per year in Indonesia. Based on studies done in six Indonesian provinces, it was estimated that in 2000, there were 37 abortions for every 1000 women between the ages of 15 and 49. The typical age of abortion-taking women is between 20 and 29 (46%), and 66% are married (Jones et al., 2010). In 2018, there were 42.5 abortions per 1000 women aged 15 to 49 on the island of Java, according to recent data conducted in Indonesia (Giorgio et al., 2020). This rate is higher than the global abortion rate, which is 39/1000 women.

According to Law No. 36 of 2009 concerning Health, women who have been raped are eligible for abortions if there are medical reasons to do so. Pregnancies that endanger the fetus' life and health, including those with severe genetic diseases that cannot be treated so that they have a negative impact on the baby and make it impossible for it to live for a very long time outside the womb, are medically indicated for medical provocatus abortions. Also, taking into account the danger to the mother's life if the pregnancy is continued.

Trisomy 13 is a serious genetic anomaly in the fetus that is one of the causes of abortion as a result of the chromosomal aneuploidy. Trisomy 13 first identified by Patau et al. in 1960 (Cammarata-Scalisi et al., 2019). Trisomy 13 affects roughly 1 in 10,000 to 20,000 live births, and more than 95% of pregnancies end in prenatal death (Satgé et al., 2017).

Due to aspiration, faulty heart architecture, and central shortness of breath, trisomy 13 congenital defects can cause infant death. The first month's mortality rate is 50%, while the first year's rate is 90%. Due to the higher prevalence of preeclampsia and the danger of maternal death associated with early birth, abnormalities in these infants also have an impact on the mother's health (Chen, 2009).

In the Criminal Code Bill it is explained that there are efforts to reform the law related to the exception setting for the criminalization of abortion, the formulation of Article 467 paragraph (2) becomes:

The provisions referred to in paragraph (1) do not apply in if a woman is a victim of the crime of rape or other crimes of sexual violence that causes pregnancy whose gestational age does not exceed 12 (twelve) weeks or has indications of a medical emergency.

There is a medical emergency condition to save the mother's life in trisomy 13 pregnancy. Termination of pregnancy avoids the adverse effects that will be experienced by babies with severe congenital abnormalities.

The problems in this paper are; (1) what are the ethical problems found in pregnancies with a fetus diagnosed with a genetic disorder due to trisomy 13? and (2) What is the procedure for implementing medical abortion provocation that fulfills ethical aspects and complies with applicable regulations

RESEARCH METHOD

This research is included in the type of normative legal research, accompanied by reports of cases of abortion. Research in the field of law is based on applicable laws and regulations. This normative legal research focuses on ethical and legal aspects of the implementation of provocative medical abortion following statutory regulations.

This normative legal research starts with articles of positive law which contain explanatory concepts regarding the limitations of the permissibility of abortions that need to be carried out due to medical emergencies. Abortions performed because there are severe abnormalities in the fetus still reap various interpretations. Saving a mother's life is a priority considering that a mother is a person who has existence and social values compared to a fetus with severe birth defects. The implementation

of safe, quality, and responsible abortion by the Health Law in Indonesia requires commitment from various parties.

The data sources used in this paper are primary data from case reports of fetal pregnancies with genetic disorders from the Gatot Soebroto Army Hospital and secondary data obtained from the literature to seek, study and collect information, concepts, theories, and related laws and regulations

RESULT AND DISCUSSION

Ethical issues in pregnancy with a fetus diagnosed with a genetic disorder due to trisomy 13

What is Trisomy 13

Trisomy 13 is a chromosomal condition with a poor prognosis that is marked by numerous severe congenital defects. Trisomy 13 is also known as Patau Syndrome because it was initially identified as a syndrome by Dr. Patau in 1960. After trisomy 21 (Down Syndrome) and trisomy 18, this is the third most common trisomy instance (Edwards Syndrome). When compared to other autosomal trisomies, trisomy 13 has the worst circumstances for congenital anomalies, psychomotor delays, mental retardation, and life expectancy (Fogu et al., 2008). Median survival is only 7-10 days in patients who are born live, and 90% live less than 1 year. The estimated mortality rate for trisomy 13 is about 50 times higher than the general neonatal mortality rate (Williams & Brady, 2019).

Neonates with trisomy 13 are generally born with low birth weight (LBW) accompanied by intrauterine growth retardation (IUGR), have one umbilical artery, prolonged persistent fetal hemoglobin, microcephaly (small head size), microphthalmia (small eye size), arrhinencephaly (without some brain tissue and skull bones), cleft lip and palate, postaxial hexdactyly, and severe cardiac and great vessel defects. In approximately 4/5 cases there is a congenital abnormality of the kidney, muscle, and bone. Most of the patients were blind and deaf, with epilepsy and severe developmental delays (Čulić et al., 2016). According to Jones (2006), there are more than 100 abnormalities that can be found in patients with trisomy 13 (Jones et al., 2010).

Antenatal Diagnosis of Trisomy 13

The initial evaluation of trisomy 13 begins with fetal nuchal translucency (FNT) performed at 11-14 weeks of gestation. This examination is to measure the ultrasound appearance of a collection of fluid under the skin behind the neck of the fetus in the first trimester of pregnancy. In fetuses with chromosomal abnormalities, heart defects, and many genetic syndromes, the thickness of the FNT is increased. The size usually appears to be greater than or equal to 3.5mm. Part of the first-trimester screening also includes measurement of the free beta subunit or total human chorionic gonadotropin (B-hCG) and pregnancy-associated plasma protein-A (PAPP-A). During the first trimester, both biomarkers appear to decline (Alzahrani, 2023).

One way to assess the presence of genetic abnormalities in the fetus is to perform a Non-Invasive Prenatal Test (NIPT) using cell-free DNA in the mother's plasma to differentiate trisomy 18 and 21 from trisomy 13.

In a retrospective study conducted by Papageorghiou (2006), it was found that ultrasound findings in fetuses with trisomy 13 during the second trimester generally are holoprosencephaly, which is an anomaly of the brain parenchymal structure due to failure of complete separation of the forebrain at the stage of brain development, small head size, facial abnormalities, heart and kidney disorders, omphalocele (exit of the abdominal cavity organs), and polydactyly. In the first trimester of pregnancies with trisomy 13, 3 major defects are easily detected: holoprosencephaly, omphalocele, and/or megacystis (very large bladder). These three clinical signs of congenital abnormalities are often found in cases of trisomy 13 and 18 chromosome abnormalities, which are found in approximately 60% of fetuses with omphalocele, in approximately 20% of fetuses with megacystis, and approximately 30% of fetuses with holoprosencephaly (Papageorghiou et al., 2006).

Risks for Mothers Containing Babies with Trisomy 13

Women carrying trisomy 13 fetuses tend to have an abnormal placenta and can experience preeclampsia in the second and third trimesters. Preeclampsia is an increase in blood pressure and excess protein in the urine that occurs after more than 20 weeks of gestation. If not treated immediately, preeclampsia can cause complications that are dangerous for the mother and fetus. If this condition is not treated immediately, it can develop into eclampsia which threatens the life of pregnant women and the fetus. This is caused by an excess of circulating proteins that dissolve in the circulation and are involved in the occurrence of preeclampsia (Jena et al., 2020).

This is caused by the presence of genes that cause preeclampsia on chromosome 13, such as sFlt1, COL4A2 and periostin. sFlt1 locks onto chromosome 13q12 and encodes a placental completion-like FMS tyrosine kinase 1 (sFlt1), which binds to vascular endothelial growth factor receptor 1 (VEGF) with high affinity. Excess circulating circulating soluble proteins such as tyrosinase kinase 1 and decreased circulating placental growth factor affect maternal circulating angiogenic proteins, which may explain the increased incidence of preeclampsia in trisomy 13 pregnancies. COL4A1 maps to 13q34 and encodes the α1 base chain of type IV collagen. Abnormal expression of the collagen IV gene can result in ineffective basement membrane remodeling and subsequent shallow trophoblastic infiltration. Periostin or OSF2 maps to chromosome 13q13.3 and encodes periostin or osteoblast-specific factor 2 (OSF2), which plays a role in the adhesion process. Release of adhesion molecules from the placenta can interfere with adhesion interactions between cells and regulate the activation of leukocytes and endothelial cells, there by stimulating inflammation (Bianchi et al., 2010).

Case Report of Pregnancy with Trisomy 13 fetus

Mrs. LD (33 years) pregnant with G2P1A0 (second pregnancy and never had an abortion), made her first antenatal visit on December 4 2022 at Brawijaya Hospital Jakarta with a gestational age of 4 weeks 5 days. The second antenatal visit was carried out on 17 December 2022 at the Brawijaya Saharjo Hospital with a gestational age of 7 weeks and 2 days, the ultrasound results showed a fetal heart rate. On January 14, 2023, at 10 weeks and 2 days of gestation, the patient's mother

was advised to perform NIPT. NIPT examination results show High Risk results on chromosome 13 or trisomy 13.

The patient was referred to the Gatot Soebroto Army Hospital, from the ultrasound results at 13 weeks 2 days of gestation, several abnormalities were found in the fetus, including clubfeet (abnormalities of legs twisted out of shape or position), omphalocele, nasal bone hypoplasia (nasal bones that do not develop), and hygroma colli (an abnormal growth on the neck). With the results of these supporting examinations, the fetus Ny. LD was diagnosed as trisomy 13 with multiple congenital abnormalities.

On the basis of poor prognosis considerations, a meeting was held by the Gatot Soebroto Army Hospital Medical Committee. The meeting was aimed at considering the feasibility of terminating the pregnancy in this patient. Families are given the option to terminate the pregnancy or continue the pregnancy with the risk that the baby will be born with severe defects and it will be difficult for the baby to survive in the long term. In addition, there is a possibility of preeclampsia which can threaten the mother's life if the pregnancy is maintained. After receiving this information, the family gave consent to terminate the pregnancy. Before termination of pregnancy counseling is carried out to patients who can be well received. Then a recommendation was issued from the Head of RSPAD Gatot Soebroto to carry out medical abortion provocation at 14 weeks of gestation which was carried out by a Fetomaternal subspecialty Obstetrics and Gynecology Specialist doctor accompanied by a Pediatrician on 6 February 2023.

From a 3-dimensional ultrasound examination at 13 weeks 2 days of pregnancy, the following picture is obtained:

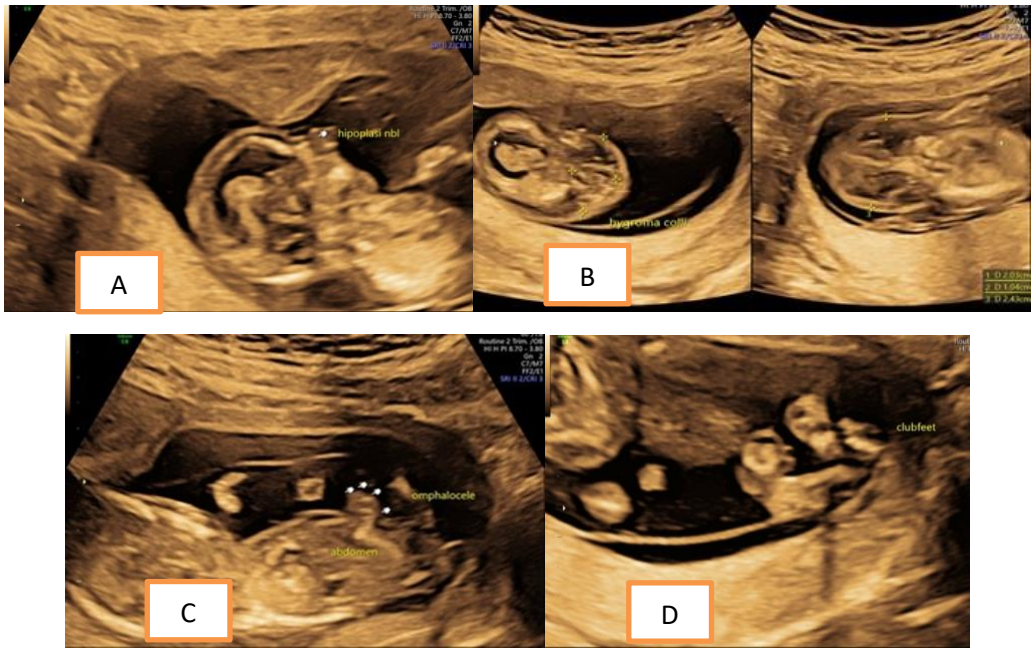

Figure 1. Ultrasound image showing congenital abnormalities A: Nasal bone hypoplasia, B: Hygroma colli (abnormal growth on the back of the neck), C: Omphalocele (discharge of abdominal contents), D: clubfeet (twisted leg)

Figure 2. After induction of labor, a fetus with a Trisomy 13 abnormality can be delivered intact (in toto)

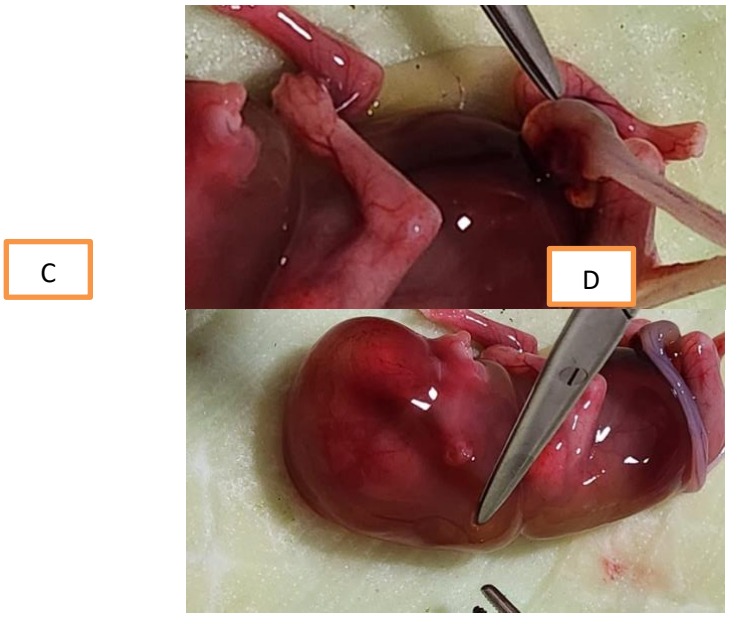

Figure 3 After the fetus is born, congenital abnormalities are obvious: A: abnormal fetal body shape, B: facial deformity, C: omphalocele, D: hygroma colli

Procedures for Implementing Medical Provocatus Abortion that Meets Ethical Aspects and Complies with Applicable Regulations

Abortion or Abortus provocatus comes from the Latin which means miscarriage on purpose. Abortus provocatus is one of the various types of abortion. If traced in the English-Indonesian dictionary, the word abortion does indeed mean abortion. In contrast to criminal abortion, namely abortion done deliberately for a reason and against the law. Abortus provocatus is a term in Latin that is officially used in the medical and legal professions. This means deliberately ending the life of the womb in the womb of a mother or pregnant woman (Seran & Setyowati, 2010).

Spontaneous abortion or miscarriage can be caused by several things, such as growth abnormalities resulting from fertilization, chromosomal abnormalities, the environment, poor content, abnormalities in the placenta, maternal diseases in the form of chronic diseases and reproductive tract disorders. Abortion is a term that is applied to all pregnancies that are terminated before the fetus is able to live outside the uterus, namely before the fetus's weight reaches 500 grams. If the weight is not known, then the gestational age can be measured, namely less than 20 weeks (139 days, counting from the first day of the last normal menstrual period) (DeCherney et al., 2019).

Analysis of Abortion in an Ethical and Legal Perspective in the Case of Trisomy 13 Fetuses

According to Bertens, there are views of pro life and pro choice. This difference in views has led to the emergence of two schools of thought that debate the issue of abortion. A pregnant woman can emphasize the right of the fetus to live. For them, abortion is the same as murder, while the pro-choice movement puts

forward the choice of whether the mother will continue her pregnancy or end it with an abortion. That is, a pregnant woman has the right to her own body, so she has the right to choose between the two possibilities, while other people in this matter cannot interfere (Bertens, 2007).

Provisions regarding abortion in medical ethics are stated in:

1) Indonesian doctor's oath: I will respect every human life from conception.

2) Article 10 KODEKI: Doctors must remember their obligation to protect the life of every human being.

According to this article, all actions and actions of doctors are aimed at maintaining the patient's health, therefore human life must be maintained with all the might. But sometimes doctors have to sacrifice one life to save another life that is more important. In the case of medical abortion provocatus (therapeutic abortion) the mother's health is sometimes a priority because of the large role of the mother in the family.

For legal reform, the Government and the DPR strengthen the regulation that the permissibility of abortions for victims of sexual violence can be up to 28 weeks or returned to 16 weeks to provide greater opportunities to build a health system that provides safe abortions.

With the promulgation of Law no. 36 of 2009 concerning Health on October 13, 2009, then Law no. 23 of 1992 concerning Health was declared no longer valid based on the Closing Provisions of Article 204 of the new Health Law. Legal arrangements regarding abortion in this Law are contained in Article 75, Article 76, Article 77 and Article 194.

Further provisions regarding indications of medical emergencies and rape, as referred to in paragraph (2) and paragraph (3) are regulated by the Government Regulation of the Republic of Indonesia No. 61 of 2014 concerning Reproductive Health.

According to Patrick (2005), there are at least 3 options that can be offered to parents with trisomy 13 pregnancies. The first option is to have an immediate abortion. Second, is early induction of labor for termination of pregnancy. While the third option is to continue the high-risk pregnancy (Guinan, 2005).

In Indonesia, the choice to have an abortion in cases of trisomy 13 is a legal action because it fulfills medical emergency indications when referring to Health Law No. 36 of 2009:

CONCLUSION

Provocatus abortion remains a controversial issue. On the one hand, they allow it for reasons that prioritize the mother's rights, while the other group believes that the child's right to life is the main thing. The legitimacy of provocative abortion in a country is very dependent on the laws in force in that country. In Indonesia, the Criminal Code states that abortion provocatus is a crime. The Criminal Code Bill has opened opportunities for medical indications and rape victims.

Termination of pregnancy in a fetus with Trisomy 13 is included in medical abortion provocatus which is carried out based on medical indications. This medical indication is based on the existence of a threat to the mother's life if the

pregnancy is continued and also the condition of the fetus that cannot live long outside the womb with severe genetic disorders.

The conclusions that can be drawn based on the explanation above are consent to termination of pregnancy with medical indications must be decided by the patient and approved by the husband/family. The purpose of this action is to prioritize patient safety in a safe, quality and responsible manner.

Termination of pregnancy in a fetus with a severe genetic disorder Trisomy 13 is a legal abortion if it is carried out according to medical indications and steps regulated in Health Law No. 36 of 2009 and Government Regulation No. 61 of 2014 concerning Reproductive Health. Doctors and medical personnel who perform legal abortions must complete the requirements and comply with regulations in order to avoid legal sanctions.

REFERENCES

Alzahrani, S., Alghamdi, M., Bin Muammar, M., Khan, A., & Tayeb, R. (2023). The Effect of Cardiac Surgery in Children with Trisomy 13 And Trisomy 18: A Systematic Review and Meta-Analysis. *International Journal Of Life Science And Pharma Research*, *13*(1).

Bearak, J., Popinchalk, A., Ganatra, B., Moller, A.-B., Tunçalp, Ö., Beavin, C., Kwok, L., & Alkema, L. (2020). Unintended pregnancy and abortion by income, region, and the legal status of abortion: estimates from a comprehensive model for 1990–2019. *The Lancet Global Health*, *8*(9), e1152–e1161.

Bertens, K. (2007). *Etika K. Bertens*. Jakarta: Gramedia.

Bianchi, D. W., Crombleholme, T. M., D'Alton, M. E., & Malone, F. (2010). *Fetology: diagnosis and management of the fetal patient*. McGraw Hill Professional.

Cammarata-Scalisi, F., Araque, D., Ramírez, R., Guaran, L., & Silva, G. Da. (2019). Trisomy 13 mosaicism. *Boletín Médico Del Hospital Infantil De México*, *76*(5), 246–250.

Chen, C.-P. (2009). Placental abnormalities and preeclampsia in trisomy 13 pregnancies. *Taiwanese Journal of Obstetrics and Gynecology*, *48*(1), 3–8.

Čulić, V., Polić, B., Mišković, S., Dragišić Ivulić, S., Žitko, V., Šipalo, T., & Pavelić, J. (2016). Patau syndrome. *Paediatria Croatica*, *60*(1), 27–29.

DeCherney, A. H., Nathan, L., Laufer, N., Roman, A. S., & Education, M.-H. (2019). *Current diagnosis & treatment: obstetrics & gynecology*. McGraw-Hill Education.

Fogu, G., Maserati, E., Cambosu, F., Moro, M. A., Poddie, F., Soro, G., Bandiera, P., Serra, G., Tusacciu, G., & Sanna, G. (2008). Patau syndrome with long survival in a case of unusual mosaic trisomy 13. *European journal of medical genetics*, *51*(4), 303–314.

Giorgio, M. M., Utomo, B., Soeharno, N., Aryanty, R. I., Stillman, M., Philbin, J., Singh, S., & Sedgh, G. (2020). Estimating the incidence of induced abortion in java, Indonesia, 2018. *International Perspectives on Sexual and Reproductive Health*, *46*, 211–222.

Guinan, P. D. (2005). Patau Syndrome and Perinatal Decision Making, Commentary 1. *AMA Journal of Ethics*, *7*(5), 336–341.

Haddad, L. B., & Nour, N. M. (2009). Unsafe abortion: unnecessary maternal mortality. *Reviews in obstetrics and gynecology*, *2*(2), 122.

Jena, M. K., Sharma, N. R., Petitt, M., Maulik, D., & Nayak, N. R. (2020). Pathogenesis of preeclampsia and therapeutic approaches targeting the placenta. *Biomolecules*, *10*(6), 953.

Jones, K. L., Jones, M. C., & Del Campo, M. (2013). *Smith's recognizable patterns of human malformation*. Elsevier Health Sciences.

Jones, R. K., Finer, L. B., & Singh, S. (2010). Characteristics of US abortion patients, 2008. *New York: Guttmacher Institute*, 20101–20108.

Papageorghiou, A. T., Avgidou, K., Spencer, K., Nix, B., & Nicolaides, K. H. (2006). Sonographic screening for trisomy 13 at 11 to 13+ 6 weeks of gestation. *American journal of obstetrics and gynecology*, *194*(2), 397–401.

Satgé, D., Nishi, M., Sirvent, N., Vekemans, M., Chenard, M., & Barnes, A. (2017). A tumor profile in Patau syndrome (trisomy 13). *American Journal of Medical Genetics Part A*, *173*(8), 2088–2096.

Seran, M., & Setyowati, A. M. W. (2010). *Dilema etika dan hukum dalam pelayanan medis*. Mandar Maju.

Williams, G. M., & Brady, R. (2019). *Patau syndrome*.