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Prevalance And Incidence Rate Of Anorectal Malformations In Newborns

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ABSTRACT
Anorectal malformations (MAR) are significant congenital abnormalities in newborns, requiring early detection and prompt treatment to prevent serious complications and improve prognosis. The aim of this study is to evaluate the prevalence and incidence rate of MAR in Indonesia, identify contributing genetic and environmental factors, and assess the effectiveness of early detection methods and treatment strategies. The research method involved literature review and reference analysis of various MAR-related studies, including epidemiological studies, case reports, and experimental research. The results of the analysis show that the prevalence of MAR in Indonesia ranges from 1 in 5,000 to 1 in 7,000 live births, with risk factors including genetic mutations, exposure to teratogenic substances, and maternal medical conditions. A multidisciplinary approach and psychosocial support are essential for optimal management and improving the prognosis of infants with MAR. In conclusion, a holistic and sustainable approach is needed to address MAR, ensure accurate diagnosis, and provide comprehensive care for affected infants and families.

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INTRODUCTION

Anorectal malformation (MAR) is a significant congenital anomaly in newborns, characterized by abnormal development of the anus and rectum. This condition requires immediate medical attention and often requires surgical intervention. MAR has a wide spectrum, ranging from mild forms, such as anal stenosis which only requires minor correction, to very complex forms, such as rectal atresia which is associated with abnormalities in other organ systems such as the genitourinary system and spine. In more complex cases, babies may require multiple surgical procedures and long-term treatment to ensure normal function. The prevalence and incidence rates of MAR vary across countries and populations, depending on genetic and environmental factors. In developed countries with good access to health, early detection and treatment may be more common, whereas in developing countries, limited health facilities can cause delays in diagnosis and intervention. Factors causing MAR include a combination of genetic factors, such as certain gene mutations, and environmental factors, such as exposure to teratogenic substances during pregnancy, maternal infections, or maternal medical conditions such as diabetes and

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obesity. Research continues to be conducted to better understand the etiology of this disorder and to develop more effective prevention and treatment methods(Islamiaty et al., 2023).

In Indonesia, data regarding the prevalence of anorectal malformations (MAR) is still limited, but several studies show that the incidence rate is quite significant and requires serious attention. Based on reports from several large hospitals in various regions, MAR is found in 1 in 5,000 to 1 in 7,000 live births. This figure is in line with global data which shows an incidence of around 1 per 4,000 to 1 per 5,000 live births. Differences in incidence rates between various studies and reports can be caused by various factors, including differences in recording methodology, availability of health facilities, and public awareness and knowledge of this disorder.(Hapsari et al., 2022).

In some areas, health facilities may not be optimal in detecting and recording MAR cases, so the actual incidence rate may be higher than reported. Apart from that, awareness of the public and health workers regarding the importance of early detection and treatment of MAR also plays a role in this variation in data. Socioeconomic factors and access to adequate health services also influence variations in reported incidence rates, where areas with better access to health services tend to have more accurate and comprehensive data. Therefore, it is important to increase efforts in more systematic data collection and education for the public and health workers to identify and treat MAR more effectively.(Matthew et al., 2021).

The causal factors of anorectal malformations (MAR) are not fully understood, but a combination of genetic and environmental factors is thought to play an important role. Several studies have shown a connection between MAR and certain gene mutations that can disrupt the normal development of the anorectal system in the fetus. In addition, exposure to teratogenic substances during pregnancy, such as certain drugs, maternal infections, and nutritional deficiencies, is also associated with an increased risk of MAR. Maternal medical conditions, such as diabetes and obesity, also increase the risk of congenital malformations, including MAR, because they can affect the intrauterine environment and fetal development. However, further research is still needed to identify the specific factors and mechanisms underlying the occurrence of MAR, in order to develop more effective prevention strategies.

The importance of early detection and treatment of anorectal malformations (MAR) cannot be ignored, considering the serious impacts that can result if this condition is not treated immediately. Babies with MAR who do not immediately receive appropriate medical treatment are at risk of experiencing serious complications such as serious infections, sepsis, and significant growth and development disorders. Therefore, early identification through careful physical examination when the baby is born is very important to detect this abnormality. A thorough physical examination may include checking for signs such as the absence of an anal canal or the presence of a fistula. In addition, the use of advanced imaging technologies such as ultrasound and MRI is helpful in more accurate diagnosis and determining appropriate intervention plans. This imaging technology allows clearer visualization of anatomical structures and potential abnormalities, so doctors can plan

appropriate surgical or other therapy procedures to ensure optimal repair and prevent further complications.

Management of MAR involves a multidisciplinary approach that includes pediatricians, pediatric surgeons, and the neonatal intensive care team. Surgical intervention is often necessary to correct anorectal anatomy and function. The surgical procedures performed vary depending on the type and severity of the malformation, ranging from simple anoplasty to complex reconstruction. Postoperative care is also important to ensure optimal recovery and prevent long-term complications. Apart from medical aspects, psychosocial support for families of babies with MAR is also important. Parents often experience stress and confusion when faced with this diagnosis. Education regarding the condition, treatment plan, and long-term prognosis is helpful in reducing anxiety and preparing families for what may be a long and challenging treatment journey.

Further research into the etiology, diagnosis, and treatment of MAR is urgently needed to improve treatment outcomes. Larger epidemiological studies may provide a more accurate picture of the prevalence and risk factors for MAR in various populations. In addition, developments in surgical techniques and postoperative management are expected to improve the quality of life of patients with MAR. Collaboration between health centers, both at the national and international levels, is also important to overcome challenges in handling MAR. Exchange of information and experience between experts can accelerate the development of knowledge and improve standards of care. In addition, the existence of a national registry for congenital malformations can help in the monitoring and evaluation of health programs aimed at preventing and treating MAR(Darussalam & Thaib, 2016).

Overall, anorectal malformation (MAR) is a complex condition and requires special attention from various medical and social aspects. Treatment of MAR not only involves surgical intervention to correct anatomical abnormalities, but also requires a multidisciplinary approach that includes support from the intensive care team, pediatric surgeons, pediatricians, and other health professionals. Apart from medical aspects, psychosocial support for families of babies with MAR is also very important, considering the stress and confusion that parents often experience when facing this condition. Adequate education and counseling can help parents understand the condition, treatment plans, and long-term prognosis, so that they are better prepared to face the challenges that arise. With a comprehensive and sustainable approach, including improvements in early diagnosis, surgical techniques, and postoperative management, it is hoped that the treatment and prognosis for babies with MAR can continue to improve, providing better hope for their future.

The aim of this study was to evaluate the prevalence and incidence rate of anorectal malformations (MAR) in newborns in Indonesia, as well as identify genetic and environmental factors that contribute to this condition. This study also aims to assess the effectiveness of early detection methods and treatment strategies currently used, with the hope of increasing understanding of MAR, improving diagnostic procedures, and developing better treatment approaches. In addition, it is hoped that this research will provide insight into the need for psychosocial support for affected families, as well as encourage increased

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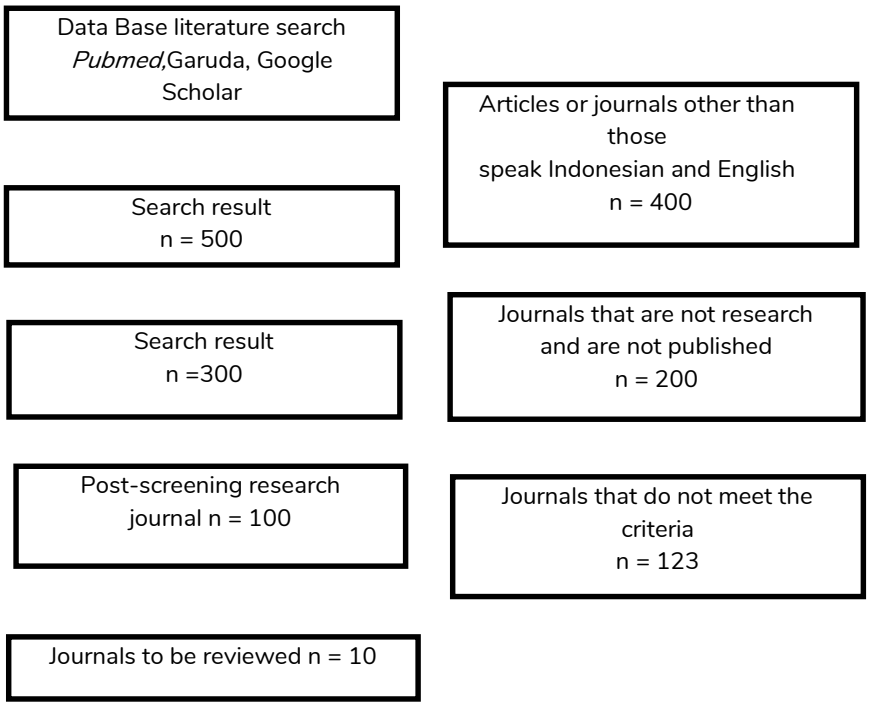
awareness and education of the public and health workers about the importance of timely identification and intervention for MAR.

METHODS

This research method involves a literature review and analysis of references from various studies that have been carried out regarding anorectal malformations (MAR). First, a literature search was conducted through major medical databases such as PubMed, Scopus, and Google Scholar using keywords such as "prevalence of MAR", "etiology of MAR", "early diagnosis of MAR", and "treatment of MAR". Selected articles included epidemiological studies, case reports, literature reviews, as well as experimental research relevant to the prevalence, causes, diagnosis, and treatment of MAR. This study also reviewed international and national clinical guidelines regarding MAR to understand best practice standards in the management of this condition.

Next, reference analysis was conducted to identify key trends and gaps in existing knowledge. Data obtained from the literature were analyzed qualitatively to identify genetic and environmental risk factors associated with MAR, as well as evaluate the most effective early detection methods and recommended treatment strategies. The study also assessed the role of imaging technologies such as ultrasound and MRI in the diagnosis of MAR, as well as the effectiveness of various surgical interventions. Through this approach, this study aims to provide a comprehensive overview of MAR, which can be used to improve clinical practice and formulate recommendations for future research.



Picture. PRISMA Flow of Research Articles Prevalence and Incidence Rate of Anorectal Malformations in Newborns

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RESULTS AND DISCUSSION

Results

In the literature screening process for this study, we started with a total of 500 initial search results. Of this number, 300 search results were eliminated because they did not meet the basic research criteria. In addition, there were 400 articles or journals that were rejected because they were written in languages other than Indonesian and English, which are priority languages to ensure readability and relevance of information. Journals that were not the result of original research or that were not published were also eliminated, with a total of 200 journals. Furthermore, 123 journals did not meet the established quality criteria, such as publication period and full text accessibility.

After going through this strict selection process, we finally selected 10 journals that will be reviewed further. These journals are selected based on research quality, relevance to the topic, and conformity to established criteria. The results of research from this literature will provide a comprehensive picture of the prevalence and incidence rate of anorectal malformations in newborn babies.By reviewing important findings from selected journals, this research is expected to identify key factors that contribute to the prevalence and incidence rate of anorectal malformations in newborns..This process ensures that the analysis carried out is based on strong and relevant evidence, thereby supporting the development of new knowledge and a deeper understanding of the prevalence and incidence rate of anorectal malformations in newborns.(Lokananta, 2015).

Analysis Results

No	Writer	Year	Article Title	Number of Samples	Research design	Results	Determinant Factors
1	Sajeel	2020	Epidemiological Comparison of Anorectal Malformation with Other Gastrointestinal Abnormalities in Pediatric Ward	150	Observational	The incidence rate of MAR is higher compared with other gastrointestinal disorders	Genetics, nutrition, maternal infections
2	N Nurlan	2023	Characteristics of Anorectal Malformation Patients	30	Descriptive	The majority of MAR patients are male, and most have additional disorders	Genetics, maternal diabetes
3	Ni Ketut Ayu Murtini	2020	Incidence of Congenital Abnormalities in Newborns According to	120	Retrospective	Congenital abnormalities including MAR are found in 10% of all live	Maternal nutrition, socioeconomic status

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No	Writer	Year	Article Title	Number of Samples	Research design	Results	Determinant Factors
			Characteristics at Sanglah Central General Hospital, Denpasar			births	
4	Insanilahia	2022	Characteristics of Anorectal Malformation Patients at Raden Mattaher Regional Hospital	180	Descriptive	MAR cases are often associated with genitourinary and spinal anomalies	Exposure to teratogens, genetic
5	Betty Anane	2023	Prevalence, Patterns, and Outcomes of Congenital Anomalies Treated in Neonatal Units in Low-Income Countries	300	Cross-sectional	Mortality rates are high in infants with MAR in low-income countries	Access to health, economic status
6	Ariadne Tiara Hapsari	2020	Predictors of Neonatal Mortality with Postoperative Anorectal Malformation	25	Prospective	The high neonatal mortality rate after MAR surgery is mainly related to sepsis	Surgical complications, nosocomial infections
7	Rabah Shwky	2019	Congenital Malformations Prevalent Among Egyptian Children and Associated Risk Factors	40	Cross-sectional	MAR is one of the common congenital abnormalities in Egypt, often accompanied by cardiovascular abnormalities	Genetics, environment, chemical exposure
8	Martono Tri Utomo	2020	Baby Girl with Apert Syndrome and Anorectal Malformation	1	Case study	A rare case of a syndrome with a combination of MAR and	Genetic

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No	Writer	Year	Article Title	Number of Samples	Research design	Results	Determinant Factors
			Without Fistula			Apert Syndrome	
9	Rudi Haryono	2020	Handling of Atresia Ani in Children	10	Descriptive	Treatment of atresia ani requires immediate surgical intervention to prevent further complications	Surgical intervention, comorbid conditions
10	Leni Ervina Khottob	2019	Factors Contributing to the Incidence of Congenital Anomalies in Neonates at Abdul Moeloek General Hospital, Lampung	35	Observational	The main risk factors for congenital anomalies including MAR are maternal medical conditions and environmental exposures	Maternal health status, environment, exposure to teratogens

Discussion

Prevalence of Anorectal Malformations in Indonesia and Globally

The prevalence of anorectal malformations (MAR) in newborns varies in various countries, including Indonesia. Available data shows that in Indonesia, the prevalence of MAR ranges from 1 in 5,000 to 1 in 7,000 live births. This figure is in line with the reported global prevalence, which is around 1 per 4,000 to 1 per 5,000 births. Variations in these figures reflect various influencing factors, both on a local and international scale. For example, differences in methods of recording and reporting cases can cause variations in reported prevalence rates. In some countries, more systematic and comprehensive recording may produce more accurate data, while in others, limitations in recording infrastructure may lead to underreporting(Haryono, 2013).

Additionally, access and quality of health facilities play an important role in determining the recorded prevalence. In developed countries, where health facilities are more accessible and of higher quality, early detection and prompt intervention for MAR is more common. This allows for quicker and more effective treatment, which in turn can influence reported prevalence rates. On the other hand, in developing countries, limited resources and health facilities can cause delays in diagnosis and treatment. As a result, many MAR cases may not be detected or receive the necessary treatment too late. The level of awareness of the public and health workers regarding this disorder also plays an important role; in areas where awareness and education regarding MAR is higher, the

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likelihood of case detection and reporting will be greater. These factors together explain why there is variation in the reported prevalence of MAR across countries and even within the same country.

Genetic and Environmental Risk Factors

A combination of genetic and environmental factors plays an important role in the occurrence of anorectal malformations (MAR). Several studies have shown a link between MAR and certain gene mutations that can disrupt the normal development of the anorectal system in the fetus. For example, mutations in genes involved in the development of the gastrointestinal tract and genitourinary system have been identified as major risk factors. Research in animals and genetic studies in humans show that mutations or impaired expression in these genes can cause malformations in the anorectal structure. Apart from that, genetic factors such as a family history of congenital abnormalities also increase the risk of MAR in subsequent offspring(Ervina et al., 2023).

Apart from genetic factors, exposure to teratogenic substances during pregnancy has a significant contribution to the risk of MAR. Teratogens, such as certain drugs (for example antiepileptic drugs or thalidomide), maternal infections (such as rubella), and nutritional deficiencies (especially folic acid), can interfere with fetal development and increase the risk of congenital malformations including MAR. Maternal medical conditions, such as diabetes and obesity, are also important risk factors. Maternal diabetes can cause hyperglycemia that affects the intrauterine environment, while obesity can cause systemic inflammation and hormonal changes that negatively impact fetal development. Although many factors have been identified, further research is still needed to identify the specific mechanisms underlying MAR. A deeper understanding of the interactions between genetic and environmental factors will help in developing more effective prevention strategies and personalizing prenatal interventions to reduce the incidence of MAR in the future(Chirul et al., 2016).

The Importance of Early Detection and Comprehensive Treatment

Early detection and timely treatment are essential in managing anorectal malformations (MAR) in newborns. Early identification through careful physical examination immediately after birth is crucial to detect signs of MAR, such as the absence of an anal canal or the presence of a fistula. This physical examination must be carried out by trained medical personnel, who can recognize the disorder early. In addition to the physical examination, the use of imaging technologies such as ultrasound and MRI plays an important role in obtaining a clearer picture of the affected anatomical structures. Ultrasound can help in evaluating the position and condition of internal organs, while MRI provides more accurate details regarding structural abnormalities. This imaging technology greatly aids in a more precise diagnosis, which then guides the planning of necessary medical and surgical interventions(Putu & Wayan, 2018).

Babies with MAR who do not receive immediate medical treatment are at risk of experiencing serious complications such as infection, sepsis and growth disorders. Therefore, a multidisciplinary approach is necessary to ensure optimal repair and prevent further complications. The treatment team should include a pediatric surgeon, pediatrician,

and neonatal intensive care team, who work together to develop a comprehensive treatment plan. In addition to medical and surgical interventions, psychosocial support for families of babies with MAR is also very important. Parents often experience stress and confusion when faced with this diagnosis, so adequate education and counseling is very necessary. Through this support, families can better understand their child's condition, long-term treatment plans, and how they can participate in the treatment process. With a comprehensive and sustainable approach, it is hoped that the prognosis for babies with MAR can continue to improve, providing hope for a healthier and better quality of life for affected babies.

CONCLUSION

In conclusion, anorectal malformations (MAR) are significant congenital abnormalities in newborns that require early detection and timely treatment to prevent serious complications and improve prognosis. The prevalence of MAR varies in various countries, including Indonesia, with genetic and environmental factors playing an important role in the occurrence of this disorder. Early identification through physical examination and imaging technologies such as ultrasound and MRI are essential for accurate diagnosis and intervention planning. A multidisciplinary approach involving pediatric surgeons, paediatricians, and the neonatal intensive care team, as well as psychosocial support for families, is necessary to ensure optimal and comprehensive care. Thus, a holistic and sustainable strategy is expected to continue to improve the prognosis and quality of life for babies with MAR.

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