WORKSHOP DETEKSI DINI KELAINAN BAWAAN DI KOTA PROBOLINGGO: MENGENAL GEJALA DAN PENYEBAB HIPOTIROID KONGENITAL PADA BAYI SEBAGAI UPAYA IMPLEMENTASI SDGS GOAL III

WORKSHOP ON EARLY DETECTION OF CONGENITAL ABNORMALITIES IN PROBOLINGGO CITY: GET TO KNOW THE SYMPTOMS AND CAUSES OF CONGENITAL HYPOTHYROIDISM IN BABIES AS AN EFFORT TO IMPLEMENT SDGS GOAL III

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ABSTRACT

The prevalence of congenital hypothyroidism in Indonesia is still not known with certainty. According to the 2007 Based Health Research Results (Riskesdas), thyroid stimulating hormone (TSH) increased 2.2% in women and 2.7% in men between 2005 and 2007. The target outcomes to be achieved are increasing awareness, interest and knowledge medical personnel to carry out early detection and management of Congenital Hypothyroidism in Probolinggo. The activity, which was held on October 1 2022 at the Bromo View Hotel, Probolinggo, was organized by the Indonesian Doctors Association Probolinggo City Branch in collaboration with the FK UNAIR Genetics Unit with the target participants being medical personnel such as doctors, nurses and midwives. Target 3 of the sustainable development agenda was partially implemented through this workshop. In addition, it is very important to immediately identify and treat congenital hypothyroidism patients based on their etiology to maximize outcomes in line with SDG goal 3, namely good health and well-being.

INTRODUCTION

The most prevalent congenital endocrine disorder in children is congenital hypothyroidism, which is also the main contributor to preventable mental retardation in children. Without early treatment, this disorder which is brought on by a deficiency in thyroid hormones in the womb can cause severe mental retardation. (Wassner, 2018).

The incidence of congenital hypothyroidism varies by country, but it is generally one in every 3000-4000 live births (Liu, 2023). Thyroid dysgenesis is the most common cause of congenital hypothyroidism, accounting for 85% of cases (Stoupa et al., 2022). The disorder affects women more frequently than men, with a 2:1.1 ratio. Compared to people of Asian descent, the incidence rate in people with black skin is very low. As a result, it is estimated that East Asia has a higher prevalence of neonatal hypothyroidism than Western countries. This CH affects 6.8 and 4.8 infants out of 10,000 in Japan and China, respectively (Pulungan et al., 2020).

There is no national data prevalence in Indonesia yet. Still, there is some data regarding Congenital Hypothyroidism in several hospitals in Indonesia, such as RSUP Dr. Cipto Mangunkusumo Jakarta from 2000 to September 2014, screening results showed 85 babies were positive 213,669 babies with a ratio of 1:2513 births. The screening results show that the numbers exceed the global prevalence of 1:3000 births. According to a medical record review at Hasan Sadikin Hospital in Bandung's endocrine clinic, 70% of those diagnosed with Congenital Hypothyroidism are over a year old. 2.3% are diagnosed at less than three months, with minimal growth and development retardation, and 70% have permanent mental retardation (Kemenkes RI. 2016).

The symptoms of Congenital Hypothyroidism in the baby's early life are very vague and atypical, and treatment delays in Congenital Hypothyroidism will result in impaired physical growth and permanent mental retardation. As a result, prevention
and early treatment with thyroid hormones can prevent physical and mental morbidity, and monitoring is required to produce treatment results and optimal child growth and development (Bin, 2023).

Therefore, these workshops' activities were designed to educate health professionals, who practiced spotting it as soon as possible, specifically when the baby was born. As goal 3 of the SDGs is implemented, early detection of congenital hypothyroidism through screening exams can also prevent children from developing intellectual disorders in the future, preventing growth disorders.

LITERATUR REVIEW

Congenital Abnormalities

Congenital or congenital disorders have been present since birth and can be caused by genetic or non-genetic factors. Congenital anomalies are birth defects, congenital anomalies, or congenital deformities. Based on pathogenesis, according to Effendi (2014), congenital abnormalities can be classified as follows:

1. Malformations
   Malformations are abnormalities caused by failure or imperfection of one or more embryogenesis processes. Some examples of malformations include cleft lip with or without cleft palate, neural tube closure defects, pyloric stenosis, spina bifida, and cardiac septum defects (Javier et al., 2023). Malformations can be classified into major and minor malformations. Major malformation is an abnormality that, if not corrected, will cause bodily dysfunction and reduce life expectancy. Meanwhile, minor malformations will not cause serious health problems and may only have a cosmetic effect. Malformations of the brain, heart, kidneys, extremities, and gastrointestinal tract are major malformations, while earlobe abnormalities, eyelid creases, finger abnormalities, indentations in the skin (dimples), extra nipples are examples of minor malformations;

2. Deformation
   Deformation is defined as the shape, condition, or abnormality of a body part caused by mechanical forces after the normal position, such as crooked legs or micrognathia (small mandibles) (Verma, R. P., 2021). This pressure can be caused by limited space in the uterus or other maternal factors such as primigravida, narrow pelvis, uterine abnormalities such as a bicornus uterus, multiple pregnancies;

3. Disruption
   Disruption is a morphological defect in one or more body parts caused by a disturbance in the initial normal development process (Kadry, H., 2020). This usually occurs after embryogenesis. In contrast to dizziness, which is only caused by mechanical pressure, the disorder can be caused by ischemia, bleeding, or adhesions. For example, strands of amniotic membrane, called amniotic bands, can detach and attach to various parts of the body, including the extremities, fingers, skull, and face;

4. Dysplasia
   Dysplasia means damage (structural abnormalities) resulting from abnormal cell function or organization, affecting one type of tissue throughout the body (Mito et al., 2022). A small percentage of these disorders involve biochemical abnormalities
in the cells, usually regarding abnormalities in enzyme production or protein synthesis. Gene mutations cause the most. Because the tissue is intrinsically abnormal, the clinical effects persist or worsen. This is different from previous pathogenesis. Malformations, mixing, and disturbances cause effects within a clear time; even though the disorders they cause may last a long time, the causes are relatively short-lived. Dysplasia can continue to cause lifelong changes.

**Congenital Hypothyroidism**

An absence of thyroid hormone at birth is known as congenital hypothyroidism (CH). The most common causes of thyroid hormone deficiency at birth are issues with thyroid gland development (dysgenesis) or poor thyroid hormone biosynthesis (dyshormonogenesis). (Moran et al., 2022). Primary hypothyroidism is brought on by this disorder. Thyroid-stimulating hormone (TSH) deficiency is the primary cause of secondary or central hypothyroidism at birth. Congenital TSH deficiency (caused by mutations in the TSH subunit gene) is rarely an isolated issue. Nevertheless, it is frequently part of congenital hypopituitarism and is linked to deficiencies in other pituitary hormones. A distinct type of hypothyroidism known as peripheral hypothyroidism is brought on by deviations in the action, metabolism, or transport of thyroid hormone.

Permanent and transient CH are the two types of congenital hypothyroidism. A lifelong course of treatment is necessary for a persistent thyroid hormone deficiency known as permanent CH (Donbaloğlu et al., 2022). A temporary lack of thyroid hormone that was identified at birth but later returned to normal thyroid hormone production is referred to as transient CH. Normal remission of euthyroidism happens during the first few months or years of life. Primary and secondary (or central) permanent CH are additional categories for permanent CH; reports of transient primary CH have also been made. Furthermore, some types of CH are characterized by harm to other organ systems; this condition is known as syndromic hypothyroidism.

Whether hypothyroidism is permanent or temporary, primary, secondary, or peripheral, as well as whether other organ systems are involved, depends on the underlying cause of CH (Kostopoulou et al., 2021). The primary CH will be the focus of this review, but secondary or central CH will also be covered in some detail. It should be noted that in many CH cases, the underlying etiology may not be known. Furthermore, only 2% of thyroid dysgenesis cases have mutations in the genes encoding transcription factors crucial for the development of the thyroid gland, even though the precise cause of some cases, such as mutations in the TTF-2 gene, is known. Thus, the precise reason for the majority of thyroid dysgenesis cases is still a mystery. However, since CH management is based on returning thyroid function to normal without knowing the precise cause, this is not a serious issue.

Congenital hypothyroidism presents with subtle early symptoms. The history of the mother and the pregnancy, however, may offer some guidance. Twenty percent of pregnancies last longer than forty-two weeks. A mother’s autoimmune thyroid disease or an iodine-deficient diet may also show symptoms. Pregnancy-related accidental radioactive iodine exposure is uncommon. Once at home, these infants are content and may sleep soundly. Constipation and hoarse crying are additional signs. More than
three weeks of neonatal hyperbilirubinemia are not uncommon. This results from the hepatic glucuronyl transferase’s infancy.

**SDGs Goal III**

Sustainable Development Goals (SDGs) is a global sustainable development agreement ratified by the United Nations (United Nations) on September 25, 2015. SDGs are global actions planned to achieve 17 goals and 169 targets for the next 15 years, from 2016 to 2030. Zero hunger (no hunger) is goal number two of the 17 SDGs. The SDGs aim to eliminate all types of hunger, including the problem of malnutrition.

Poverty can leave generations vulnerable to disease and malnutrition and unable to enjoy education, so poverty will bequeath generations of people with social problems. That is why poverty will ultimately become a burden on the state and society to this day.

The goal of a country is development, where a country will become more advanced if it experiences an increase in its development. One indicator of development success is increasing high economic growth, so it is hoped that it will overcome unemployment and poverty.

Because of the complexity and dynamic nature of human interactions with the environment and the relationships between components in bio-physical, technical, and human systems, it can be difficult for people to find secure environments in which to grow. This makes room for important sector education to equip young people with the knowledge and abilities to address current and upcoming global challenges like climate change, digitalization, and globalization. Young people need to receive an education that takes into account the realities of these challenges because the world is interconnected, dynamic, and complex, with global challenges. The need for education that supports a more sustainable world is becoming more obvious because education can enhance people's lives and contribute to sustainable development. Sustainable development aims to integrate economic development with environmental concerns and social integrity, fairly sharing costs and benefits to support the wellbeing of the current generation without jeopardizing the needs of future generations.

SDG III aims to ensure healthy lives and improve welfare for all in society. All health-related issues are covered in SDG number 3 which aims to ensure healthy lives and improve well-being for all people at all ages. There are 38 SDG targets that must be met by the health sector. Apart from problems that have not been completely resolved, new problems have emerged. Examples include programs to reduce maternal and infant mortality rates (MMR and IMR), control HIV/AIDS, tuberculosis and malaria, and increase access to reproductive health (including family planning).

All stakeholders, including central and regional governments, parliament, business, media, social institutions, professional and academic organizations, and development partners, must actively participate in health sector development for the SDGs.

Combining the concepts of integrated development and health into a number of development management processes, such as input, process, output, outcome and development impact, is the most challenging aspect in implementing the sustainable development agenda in Indonesia.
To achieve the SDGs in the area of health, the Healthy Indonesia Program is being promoted. It has three pillars: a) In order to improve access and quality of care, b) Health services are carried out and directed in accordance with the healthy paradigm, which prioritizes promotive and preventive concepts. With the aid of clinical governance, management governance, and program governance, primary health services are concentrated on promotive and preventive service efforts. And c) National Health Insurance: The government is committed to ensuring access to healthcare for all Indonesian citizens and visitors.

**METHOD**

Direct counseling is provided as part of this community service project at the Bromo View Hotel in Probolinggo, which was organized by the Probolinggo City branch of the Indonesian Doctors Association in association with the Genetics Unit of the Faculty of Health Airlangga University (FK UNAIR). One hundred health professionals from various fields in Probolinggo City attended this workshop, including general practitioners, general pediatricians, midwives, nurses, and medical students. On Saturday, October 1, 2022, starting at 9 AM, the educational method was implemented through free webinars with public lectures, educational videos about Congenital Hypothyroidism (CH), and a Q&A session. Free credit vouchers are offered to the first 100 registrants to pique interest among participants. The topics covered included managing CH diagnosis and treatment, early detection of CH, and several case simulations. There were also workshops with other materials, including congenital deafness, differences in sex development, and genetic examination of congenital abnormalities.

**RESULT AND DISCUSSION**

There are five target participants in this Workshop on Early Detection of Congenital Abnormalities, such as general practitioners, general pediatricians, midwives, nurses, and medical students from various fields in Probolinggo City.

We obtained data from the Indonesian Doctors Association Probolinggo City Branch that the number of Congenital Hypothyroidism cases in Probolinggo City is relatively high and has not been handled well. As a result, the FK UNAIR Genetics Unit offered an educational strategy that included playing educational videos about Congenital Hypothyroidism and conducting free webinars with public lectures (CH) and Q&A by a pediatric and genetic consultant (Figure 1) to raise awareness, interest, and knowledge of the participants.
According to Setyaningsih et al. (2022), many healthcare professionals still lack blood collection training to screen for congenital hypothyroidism. Thus, it is often too late to properly diagnose and treat many cases of congenital hypothyroidism. Due to this, many healthcare professionals occasionally adopt a defensive attitude. When performing their responsibilities as CHSP implementation specialists, they experience anxiety (Rayhan et al., 2022).

Regarding facilities and infrastructure, only a limited number of filter paper and lancet blood sample collection facilities are available, making it impossible to implement the CHSP if targets are identified, but facilities are not. The absence of complete facilities for educating healthcare professionals on recognizing the signs and causes of congenital hypothyroidism is also regarded as a lack of supporting facilities for activities such as brochures, posters, and educational videos (Noflidaputri et al., 2021). Contrary to the study's findings, which asserted that the CHSP's distribution of supporting facilities had gone smoothly, by Anggraini et al. (2019).

In terms of time management, there are four important things to consider, such as:

1. **Congenital hypothyroidism is often detected too late.**

   Congenital hypothyroidism usually doesn't show any symptoms until several months later. We should be aware of the various signs and causes of congenital hypothyroidism in infants, such as swollen face, dry, thick baby skin, difficulty eating, and typically, trouble drinking. Ninety-five percent of newborns with congenital hypothyroidism exhibit no symptoms at birth, and even those that do are ambiguous and untypical (Anton, 2020).

2. **Delay in diagnosis will impact the child's physical and mental maturity**

   Thyroid hormones play an important role in brain development, and perinatal thyroid hormone deficiency results in severe developmental delays. Perinatal thyroid hormone deficiency is clinically known as congenital hypothyroidism, caused by thyroid gland dysgenesis or a lack of iodine. Thyroid hormone deficiency disrupts neuronal architecture, and neuropathological findings such as abnormal synapse formation, defects in neuronal migration, and impairment of myelination are observed in the brains of such patients if the disorder is not diagnosed or treated early (Uchida, 2021).
Figure 2. Flowchart for newborn screening to identify congenital hypothyroidism

Serial Newborn Screening Increases Congenital Hypothyroidism Detection in Preterm Infants

The efforts made will be more effective if you identify a child with congenital hypothyroidism as soon as possible. The child’s IQ development will also be influenced by the age at which therapy begins. Because of this, screening is crucial. The best time to recommend screening for congenital hypothyroidism is between 48 and 72 hours after birth. Congenital hypothyroidism requires lifelong treatment in infants (Kaluarachchi, 2019).

Positive screening outcomes on filter paper are required (TSH 20 mU/L) with serum blood before beginning treatment. If the serum FT4 level is low, treatment must start immediately. For additional assessment and treatment, suspect laboratory results (high TSH but normal FT4) must be referred to a specialized consultant endocrinologist (Bowden, 2023).

Adequate treatments for babies with congenital hypothyroidism

Levothyroxine is administered as soon as the diagnosis of congenital hypothyroidism is made. It is ideal to administer medication before the infant turns two weeks old. Levothyroxine is first administered at a dose of 10-15 g/kgBW/day, and then the dosage is changed based on age, the results of routine TSH and fT4 tests, and other factors. Congenital hypothyroidism in children necessitates lifelong treatment. The dosage of hypothyroid medication is adjusted to thyroid hormone levels in babies and children. After taking the medication, the child’s thyroid will be checked regularly every few months (Yati et al., 2017).

Implementation of SDG Goal 3 (Good Health and Well-Being)

Good health and well-being are one of the 17 goals in the sustainable development agenda that must be achieved. Good health and welfare are sustainable development goals related to the welfare and health of society (Sarma, 2023).
1) The third Sustainable Development Goal (SDG) has nine targets, one of which is to reduce the MMR to 70 per 100,000 KH or less; 2) Put an end to preventable infant and child deaths; 3) Put an end to AIDS pandemic; 4) Eliminate malaria, tuberculosis, and other tropical diseases; 5) Cut the number of premature deaths brought on by non-communicable diseases by one-third; 6) Improve the efficiency of drug abuse prevention and treatment; 7) Decrease the number of deaths and injuries from automobile accidents; 8) Ensure that everyone has access to sexual and reproductive health services; 9) Achieve universal health coverage; 10) Manage emergencies and crises. The Workshop on Early Detection of Congenital Abnormalities is targeting number 8 of these; and 11) According to the WHO, having this understanding ensures access to health services that are promotional, preventative, curative, and rehabilitative.

CONCLUSION

Delay in treatment of congenital hypothyroidism will lead to impaired physical growth and long-term mental retardation because the early symptoms of congenital hypothyroidism are very vague and non-specific. Thyroid hormone treatment can, therefore, be used to prevent both physical and mental suffering, and monitoring is necessary to ensure that treatment is working as intended and that children are growing and developing normally. By holding this activity, it is also hoped that the death rate and mortality due to congenital abnormalities can be reduced as low as possible. Additionally, this workshop program supports SDG goal 3, promoting good health and well-being.

REFERENCES


