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Case Report
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Bronchopneumonia and Multiorgan Dysfunction in a 10-month-old Infant with Down Syndrome

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Abstract

Background: Common chromosomal disorders like Down Syndrome (DS) are linked to a high rate of congenital abnormalities, especially hypothyroidism and congenital heart disease (CHD).

Presentation Case: This case report presents a 10-month-old female with DS who was brought to the emergency department with respiratory distress, poor weight gain, and delayed development. Physical examination revealed characteristic dysmorphic features, hypotonia, and a systolic murmur. Investigations showed elevated TSH and FT4, consistent with congenital hypothyroidism, while echocardiography confirmed the presence of ASD.

Discussion: This case illustrates the multisystem involvement commonly observed in Down syndrome, including congenital hypothyroidism and atrial septal defect, both of which are major contributors to morbidity and mortality. The co-occurrence of recurrent respiratory infections, heart disease, and endocrine dysfunction underscores the need for early screening, multidisciplinary management, and comprehensive care to improve outcomes in children with Down syndrome.

Conclusion: Early detection of CHD and hypothyroidism in children with Down syndrome is essential to prevent complications and support optimal growth and development through timely, multidisciplinary intervention.

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INTRODUCTION

Trisomy of chromosome 21 causes Down Syndrome (DS), the most prevalent genetic condition that causes intellectual disability. DS manifests with multi-system involvement, primarily affecting the musculoskeletal, neurological, and cardiovascular systems, with congenital heart disease (CHD), especially atrioventricular septal defect (AVSD), being most prevalent. Individuals with DS are also susceptible to hypothyroidism, autoimmune disorders, epilepsy, sensory impairments, hematologic abnormalities, growth delays, and immunodeficiency leading to recurrent infections[1,2]. The global prevalence of DS is approximately 1 in 1,000 to 1 in 1,100 live births, with an estimated 3,000 to 5,000 affected births annually, according to the World Health Organization. Riskesdas data showed DS prevalence at 0.12% in 2010, increasing to 0.13% in 2013, and rising again to 0.21% in 2018[3].

A key risk factor is advanced maternal age, with incidence rising significantly after age 35[4(4)]. Genetic contributions include parental chromosomal translocations and a family history of DS; however, most cases are sporadic and result from nondisjunction during gametogenesis. Environmental factors such as exposure to harmful substances, maternal health status, smoking, folic acid supplementation, and use of oral contraceptives may also influence the risk[5,6]

More than 50% of individuals with DS present with CHD, with AVSD (42%), ventricular septal defect (22%), and atrial septal defect (16%) as common types[7]. Endocrinopathies, particularly congenital hypothyroidism, occur in 25.3%–60% of newborns with DS, who typically exhibit elevated TSH and reduced thyroxine levels[8,9]. Early recognition of clinical signs and implementation of appropriate screening can facilitate prompt intervention, minimizing complications and reducing mortality. This report focuses on the risk factors and multiorgan screening relevant to a child with Down syndrome.

CASE PRESENTATION

A 10-month-old infant with clinical features of Down syndrome presented with complaints of shortness of breath for the past 2 days. Before this, the patient had a cough, a runny nose, and a fever for 3 days. Shortness of breath worsened with crying. The patient became irritable and refused to eat or drink. The mother noticed that the baby appeared increasingly weak.

The patient had previously experienced shortness of breath and cyanosis while breastfeeding and crying at the age of 2 weeks, which led to admission in the NICU for 18 days. After that episode, the patient had no further complaints of dyspnea. There was no history of chronic cough or frequent illness.

Birth history revealed delivery by cesarean section at full term with a birth weight of 2.3 kg and a length of 43 cm. The baby did not cry immediately after birth. Antenatal care (ANC) was conducted monthly with a midwife. At 9 months of pregnancy, decreased fetal movement was noted, prompting a cesarean delivery. The mother regularly took folic acid throughout pregnancy. There was no history of preeclampsia, gestational diabetes, or TORCH infection, but she was frequently

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exposed to cigarette smoke from her husband. The pregnancy occurred at the age of 24 years.

The patient is the second child in the family; the first child is healthy and does not have Down syndrome. The mother was 19 years old at the time of her first pregnancy. There is no history of miscarriage or Down syndrome in the family.

Developmental delays were observed: the patient cannot stand while holding on or sit without support. The baby has been able to roll over and lie prone since 8 months of age, but is unable to crawl. The patient does not yet smile spontaneously or engage in interactive play. For over a month, the baby has been able to make eye contact when spoken to and respond to sound, but has not yet begun babbling.

The baby has been given complementary foods since 6 months, consuming filtered porridge. Weight gain has been around 0.5 kg per month. Immunization history includes: Hepatitis B, BCG, oral polio (1, 2, 3), DPT-HB-HIB (1 and 2), rotavirus, and PCV (1 and 2).

Physical examination revealed:

- Weight: 4.5 kg; Length: 58 cm; Head circumference: 37 cm
- Signs of dyspnea, facial characteristics consistent with Down syndrome: round face, upslanted palpebral fissures with epicanthal folds, small flat nose, small mouth with protruding tongue
- Thorax: suprasternal retractions and fine wet crackles in the right lung
- Extremities: warm, dry, reddish acral areas with CRT <2 seconds, short fingers, and a gap between the first and second toes

Laboratory results:

- Hemoglobin: 11.0 g/dL, Leukocytes: 8,930 /µL, Platelets: 56,000 /µL, PCV: 31.4%, Neutrophils: 3,270 /µL, Lymphocytes: 5,120 /µL, NLR: 0.64
- TSH: 10.06 µIU/mL, T3: 0.87 ng/mL

Chest X-ray showed patchy infiltrates in the perihilar region of both lungs.

Echocardiography findings: Atrial septal defect (ASD) secundum, with a gap measuring 37–42 mm, indicating moderate left-to-right shunt; Severe tricuspid regurgitation (TR) with high probability of pulmonary hypertension; Right atrium (RA) and right ventricle (RV) dilatation

Management: IV fluid: Kaen 1B 450 cc/24h; Oxygen: 2 L/min via nasal cannula; Antibiotics: IV ampicillin 2 × 225 mg, IV gentamicin 1 × 50 mg; Analgesic-antipyretic: IV sodium metamizole 3 × 50 mg; Nebulization: Salbutamol ½ ampule twice daily; Hypothyroidism therapy: Euthyrox 1 × 50 mcg

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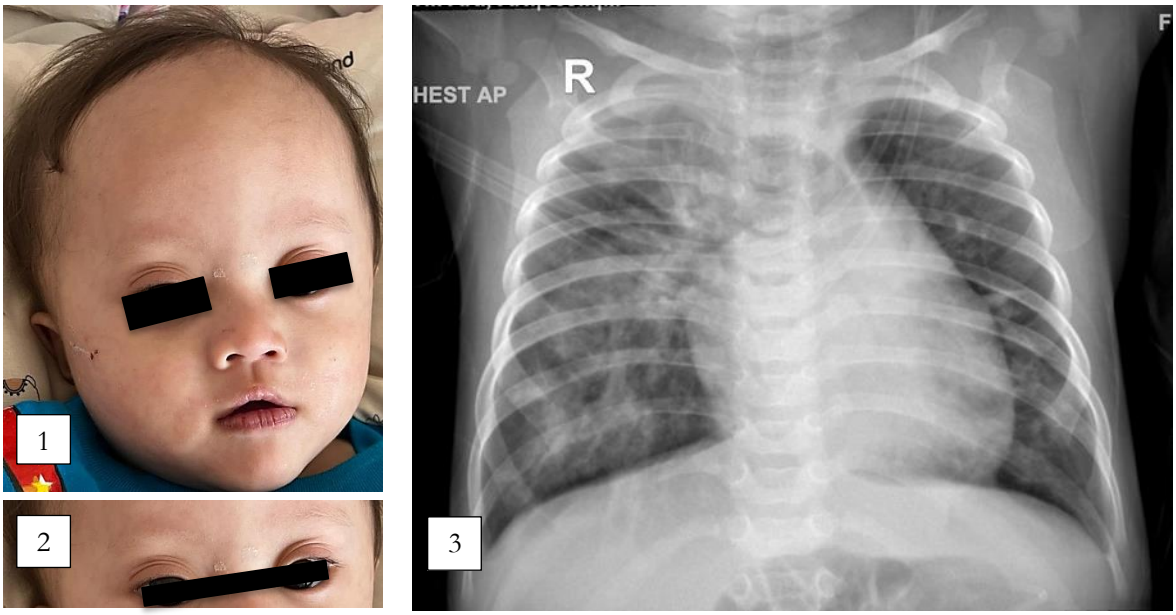

Figure 1. Mongoloid Face, 2. Epicanthal Fold, 3 Chest X-Ray PA (Source: Dwi,2024).

DISCUSSION

This case underscores the clinical complexity and characteristic multiorgan involvement frequently encountered in individuals with Down Syndrome (DS). The coexistence of congenital hypothyroidism and atrial septal defect in this patient aligns with the well-established spectrum of comorbidities associated with trisomy-21. DS is recognized as an achromosomal anomaly marked by a high prevalence of multisystem disorders, particularly congenital heart disease (CHD) and thyroid dysfunction. Both conditions, as absorbed in this case, represent critical contributors to morbidity and mortality among affected individuals and highlight the importance of early identification and integrated management strategies[1].

CHD occurs in more than 50% of DS cases and contributes significantly to early morbidity and mortality[7]. The epidemiology of congenital heart disease (CHD) in children with Down syndrome in Indonesia shows that the prevalence of CHD is quite high in this population. Based on clinical data and local studies, the prevalence of CHD in children with Down syndrome in Indonesia ranges from 40% to 60% [9,10]. The most common types include atrioventricular septal defect (AVSD), ventricular septal defect (VSD), and atrial septal defect (ASD). In our case, echocardiography examination revealed a secundum ASD with left-to-right shunting, tricuspid regurgitation, right heart dilatation, and signs of pulmonary hypertension. These findings underscore the importance of routine early echocardiography, as recommended by the AAP and DSMIG [9,11]. Recurrent respiratory infections in this patient are likely exacerbated by both pulmonary overcirculation and the immunologic susceptibility seen in DS[1,2].

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Congenital hypothyroidism is another frequent and clinically relevant condition in children with DS, with a prevalence ranging from 25% to 60%[12]. The prevalence of hypothyroidism in patients with DS is 28 times greater than that of the general population in Indonesia. In a study conducted by the Faculty of Medicine at Gadjah Mada University (UGM) involving 33 patients with Down syndrome, 27 of them were found to have hypothyroidism[13]. In this patient, markedly elevated TSH and low FT4 confirmed the diagnosis. The pathogenesis may include thyroid dysgenesis, delayed maturation, or autoimmune thyroiditis, all of which are associated with immune dysregulation in trisomy 21 [14–16]. Early diagnosis and treatment with levothyroxine are crucial to avoid neurodevelopmental impairment[17].

Advanced maternal age is a well-established risk factor for DS. In this case, the mother was 24 years old during pregnancy, with no history of children or of having Down syndrome. The risk of meiotic nondisjunction increases significantly after age 35 due to aging-related cellular and chromosomal factors [5,18,19]. Additionally, folic acid supplementation during pregnancy has been linked to reduced risk of chromosomal abnormalities and congenital heart defects. The absence of folic acid intake, as in this case, may have contributed to both the chromosomal anomaly and the heart defect. Polymorphisms in folate-metabolism genes, such as MTHFR and MTRR, have also been associated with a higher risk of DS and CHD [20–22].

This case emphasizes the interconnectedness of maternal factors, genetic susceptibility, and multiorgan complications in DS. Comprehensive screening protocols and multidisciplinary care are necessary for early diagnosis, timely intervention, and better long-term outcomes.

CONCLUSION

Children with DS are at a substantial risk of developing multiorgan complications, including hypothyroidism and congenital heart disease. Maternal factors, such as age and folic acid supplementation, remain crucial for risk mitigation. Early identification of congenital comorbidities and multidisciplinary management are essential to improve outcomes in children with Down syndrome.

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