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# Review of Infant Endocrine System Conditions Related to Coronary Abnormalities

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**Abstract**

Babies with hormonal imbalances exhibit characteristic dysmorphic symptoms and suffer from neuropsychiatric, cardiovascular, gastrointestinal, eye problems, deafness, endocrine disorders, blood disorders, and many other health problems. It's easy. Maybe so. We present a case of a newborn with Down syndrome. The baby was a full-term woman who delivered by caesarean section. She was diagnosed with a complex congenital malformation before she was born. By the 10th day of life, she began experiencing dyspnea, persistent respiratory acidosis, and persistent severe hyponatremia, requiring intubation and mechanical ventilation. Her condition was deteriorating rapidly, so our team decided to test her for metabolic disorders. Screening was positive for heterozygous Duarte-mutated galactosemia. Further tests were performed for metabolic and endocrine problems that may be related to hormonal imbalance, leading to the diagnosis of hypoaldosteronism and hypothyroidism. This case was a challenge for our team as the infant also exhibited multiple metabolic and hormonal deficiencies. In addition to congenital heart defects, neonates with hormonal imbalances may suffer from metabolic and hormonal deficiencies that can adversely affect prognosis in the short and long term. Therefore, interdisciplinary teams are often required.

**Keywords:** Hormone disorders; Newborn; Galactosemia; Hypothyroidism; Multidisciplinary team

## Introduction

Hormone disorders are among the most common chromosomal abnormalities diagnosed in newborns. It is also called trisomy 21 because it is caused by the presence of the 21st chromosome. According to a recent study, in 2015 he had 419,000 people with Down syndrome in Europe. Patients with this syndrome can suffer from multiple medical conditions, ranging from intellectual disability to congenital heart disease, celiac disease, and endocrine disorders [1].

Studies have shown that the incidence of congenital heart disease related to hormonal imbalance is approximately 45-50% of people with this syndrome. The most common cardiac defects occurring prenatally in fetuses with hormonal imbalances are atrioventricular septal defects, ventricular septal defects, secondary atrial septal defects, and patent ductus arteriosus [2]. Endocrine disorders such as obesity, diabetes, short stature, vitamin D deficiency, and thyroid dysfunction are also common in people with hormonal imbalance. Patients with hormonal disorders have significantly improved life expectancy. Life expectancy in the United States in the 1950s was 4 years. 58 years have passed in 2010. Medical care of newborns diagnosed with hormonal abnormalities requires a multidisciplinary team. The long-term and short-term prognosis of these patients is improved by reliable screening programs that help identify relevant malformations as early as possible [3].

## Case

We present the case of a full-term girl. Her birth weight was 3990 g and she was born at a gestational age of 38-39 weeks (postmenstrual gestational age) [4]. Her pregnancy was checked by an obstetrician and gynecologist. This pregnancy was a high-risk pregnancy as her mother had previously undergone a caesarean section and her fetus had been diagnosed with a heart defect in the uterus [5]. She was born by cesarean section with an Apgar score of 8 for one minute. Immediately after birth, the newborn showed a generally satisfactory appearance. She had facial features characteristic of Down syndrome, no heart murmurs, and no other overt signs of stress. Her medical team knew she had a congenital heart defect, so she was admitted to the neonatal intensive care unit (NICU).

A cardiologist diagnosed the patient with a complete atrioventricular defect (Lastelli type C, left and right atrioventricular dysfunction grade II, small functional atria) with a large atrial septal defect. She was put on medication with furosemide, spironolactone and captopril, and she tolerated the disease well and began to thrive. On the second day after birth, a grade II/VI systolic murmur was heard on cardiac auscultation [6].

At 7 days of age, the infant was well, with mild jaundice, respiratory status was stable, and her SpO2 in the air was 96% of hers and remained stable. The neonatal care team was able to initiate enteral feeding with good digestive tolerance. She was bottle-fed with artificial milk (partially hydrolysed milk). She lost 10% of her weight. Blood tests, abdominal and transwell ultrasounds were within normal limits. Her general appearance changed on her ninth day of life. She had a sudden increase in jaundice (transcutaneous bilirubin 13.8 mg/dl), her systolic murmur increased (grade III/VI), and she lost weight. She began to require 40% inspired oxygen (FiO2) to maintain her SpO2 above her 95%, with signs of dyspnea (intercostal and subcostal retraction, frequency of approximately 70 breaths/min). was also shown [7]. She also developed 100 percent bradycardia when her ventricular heart rate decrease. From that moment on, she required continuous oxygen supplementation (both invasive and non-invasive respiratory support) and respiratory stimulation (exercise therapy) [8].

Because of his rapid and sudden deterioration, the medical team

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decided to hire a metabolic disease specialist. Results showed elevated galactose levels. Studies continued in this direction by administration of galactose-1-phosphate uridylyltransferase, confirming the diagnosis of heterozygous galactosemia or homozygous Duarte-2 mutant. First, the patient was given special galactosemia milk (soybean-based), and after 24 hours his respiratory function improved so much that he was able to go off the ventilator [9].

## Discussion

Down syndrome is not only the most commonly identified chromosomal abnormality, but also one of the most studied in the last 150 years. The most common malformations associated with hormonal imbalances are cardiac, gastrointestinal, musculoskeletal, urinary tract, and endocrine malformations. In this case, a congenital heart malformation was diagnosed prenatally and required admission to a stage III maternity ward equipped with a state-of-the-art neonatal intensive care unit and multidisciplinary team [10].

As previously mentioned, this patient suffered from shortness of breath for most of her hospital stay. This led the medical team to make several differential diagnoses, including pneumonia, pulmonary edema, and ear, nose and throat (ENT) disease. Detecting the heart defect within 24 hours of birth enabled the medical team to begin special treatment and long-term monitoring as soon as possible. Her shortness of breath had several causes. First, complex cardiac abnormalities, pulmonary hypertension secondary to cardiac abnormalities, narrowing of the upper airway typical of Down's syndrome, gastrointestinal reflux, metabolic and endocrine disorders.

Galactosemia is an inherited metabolic disorder that affects carbohydrate metabolism. Severe galactose-1-phosphate uridylyltransferase (GALT) deficiency and classic galactosemia can be fatal in neonates. Certain variants of galactosemia exist. Duarte mutants are characterized by GALT activity in approximately 50% of erythrocytes (when homozygous). Duarte mutation heterozygotes have approximately 75% GALT activity. While classical galactosemia is considered a medical emergency, the Duarte variant is considered asymptomatic and does not usually require a low-galactose diet. Our patient had no gastrointestinal symptoms typical of typical galactosemia (good digestive tolerance, no loose stools or vomiting). Her blood count showed atypical respiratory acidosis of galactosemia rather than metabolic acidosis. However, starting a galactose-restricted diet with a soy-based formula significantly improved shortness of breath and respiratory acidosis.

Hypoaldosteronism is an endocrine disorder characterized by aldosterone deficiency or defective aldosterone activity at the tissue level. Severity is usually inversely proportional to age. Despite adequate intravenous correction, persistent hyponatremia was suspected in our patient. I decided to test my serum cortisol (in normal range) and serum aldosterone. Therefore, oral treatment with fludrocortisone was initiated.

## Conclusion

In summary, the cases we present highlight the need for multidisciplinary teams to deal with such complex cases. Endocrine dysfunction has a significant impact on development in this patient, underscoring the importance of screening for endocrine dysfunction in Down syndrome patients, even if the initial assessment was normal.

All newborns have the right to comprehensive medical care, regardless of the severity of their illness or prognosis. The parents of this newborn were aware of the seriousness of the disease even before birth and made the decision to continue the pregnancy, and to the best of our knowledge, have not regretted that decision.

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