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Case Report

Low Oxygen Saturation Levels Without Cardiopulmonary Pathology: Identifying A Rare Hemoglobinopathy (Hb Titusville) - A Case Report

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Introduction

A rare hemoglobinopathy was found in a female newborn with low oxygen saturation without evidence of cardiopulmonary pathology.

The detected hemoglobin, called Hb Titusville, is rarely described in literature. A characteristic property of Hb Titusville is a reduced oxygen affinity. The correct diagnosis may avoid unnecessary examinations and interventions.

Keywords: Low oxygen saturation level; Hb Titusville; Hemoglobinopathy; Oximetry; Case report

Case Report

A rarely described hemoglobinopathy rarely described in literature was found in a female newborn with low oxygen saturation levels.

A female newborn was delivered at a gestational age of 40 + 3 weeks. A few hours after birth, it was admitted to the neonatal intensive care unit due to presumed respiratory maladaptation.

Clinical Findings

The chest X-ray showed no pathological findings. Bacterial and viral infections were ruled out. Echocardiography revealed normal findings. Despite the child's clinically stable condition, the oxygen saturation was measured as to be only 90-92% by pulse oximetry (SpO2).

The mother subsequently reported that low SpO2 readings had also been detected in her own case. Her apparently healthy brother also had been found to show low SpO2 readings. He had undergone extensive cardiopulmonary examinations, yielding normal results.

Diagnostic Assessent

The SpO2 measurement of the newborn was confirmed in an arterial blood sample. At a pO2 of 100 mmHg O2 saturation was only 91%. The P50 (paO2 at which half of the hemoglobin is oxygenated) was 48 mmHg (normal range newborns: 18-24 mmHg; lower due to fetal Hb levels).

Before genetic testing, a high performance liquid chromatography, HPLC, was performed, with detection of an aberrant hemoglobin. In the genetic analysis Hb Titusville was identified. This hemoglobin has a reduced oxygen affinity.

Discussion

In pediatric medicine, hemoglobin anomalies must be included in the diagnosis as a possible underlying cause of low oxygen saturation without any clinical findings [1].

Hemoglobin Titusville in this case is a rare, inherited defect of hemoglobin structure that is most commonly found in individuals

of northern European background, which was not the case in our patient [2].

Low oxygen affinity Hb variants do not require any specific management. However, in individuals with low SpO2, the correct diagnosis is important to avoid unnecessary cardiorespiratory examinations and interventions [3]. Family history is a mainstay of diagnosis.

Authorship Contribution Statement

Mascha Schoenfeld: Writing: original draft, visualization, conceptualization.

Magnus Günther, Julia Winter, Friederike Häuser, Eva Mildenberger: Writing: review & editing, validation, supervision.

Declaration of Competing Interest

The authors declare no conflicts of interest.

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