ORIGINAL ARTICLE

# Diagnosis and treatment of neonatal machete syndrome

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## ABSTRACT

**Aim:** To provide suggestions for clinical diagnosis, treatment and eugenics through the retrospective analysis of a case of neonatal machete syndrome.
**Methods:** The clinical data of a newborn with machete syndrome admitted to the pediatrics department of Baogang Hospital of Inner Mongolia were analyzed, and suggestions of eugenics were provided based on the literatures.
**Results:** The child, male, natural conception, G2P2, gestational age 38+4 weeks, was born by cesarean section (at am 09:24 on November $13^{th}$, 2021). He was hospitalized in our department for 9 days and given NCPAP positive pressure ventilation to give oxygen and anti-infection symptomatic treatment. The dyspnea of the child was not significantly alleviated, and the heart murmurs became increasingly obvious. The child was transferred to the neonatology Department of the Seventh Medical Center of the PLA General Hospital for further treatment on 9 days after birth. After admission, piperacillin and tazobactam were treated with anti-infection, high-frequency oscillation mode of invasive ventilator, limited fluid volume and other symptomatic treatments. Fiberbronchoscopy showed atelectasis of right lung and tracheomalacia of left lung. Echocardiography showed partial anomalous pulmonary venous drainage, atrial septal defect (secondary foramina), inferior vena cava dilatation, pulmonary hypertension, initial coronary artery dilatation, and patent ductus arteriosus. CT 3D imaging of pulmonary veins showed: Congenital heart disease: partial anomalous pulmonary venous drainage (subcardial), consideration of "machete syndrome," atrial septal defect, consideration of pulmonary hypertension, right pulmonary artery stenosis, right lower lung receiving abdominal aorta blood supply, right lung tissue dysplasia, and widening of inferior vena cava pulmonary veins. Therefore, the comprehensive diagnosis was "machete syndrome." Considering the high surgical risk of the child, conservative symptomatic treatment was recommended, and the family requested discharge after 22 days of treatment. The patient died at home at 3 months follow-up after discharge.
**Conclusions:** Machete syndrome is a rare congenital property of abnormal partial or complete pulmonary venous drainage from the right or left lung to the inferior vena cava. With an incidence of approximately 2 in 100,000 live births, the syndrome is commonly associated with right lung dysplasia, pulmonary sequestration, persistent left superior vena cava, and right tilt of the heart. In this case, the child had these syndromes after birth, since the primary hospital did not recognize the disease, the child was clearly diagnosed in the higher hospital. Children with machete syndrome can be treated with surgery in the early neonatal period, but the indication of surgical treatment is still controversial, and the need for surgery is evaluated according to the severity of the patient's symptoms and pulmonary circulation blood flow.

**Key Words:** Newborn, Machete syndrome

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## 1. DATA AND METHODS

### 1.1 General information

The child, male, 4 hours after birth, the second fetus, was born in our hospital by cesarean section due to cicatricial uterus at 38+4 weeks gestational age (at am 9:24 on November $13^{th}$, 2021). The regular birth test during pregnancy showed no abnormality. His birth weight was 3,450g. The Apgar score was 10 in 1, 5 and 10 minutes. He was transferred to the NICU after groaning and spitting shortly after birth. Admission for physical examination: Body temperature 36.0°C, pulse 145 times/min, breathing 55 times/min, blood pressure 67/36 mmHg. The child showed full-term appearance, the whole body skin was slightly pale, with slightly blue around the mouth, no tumor and no hematoma. The fontanelle is flat and soft, about 2.0 cm × 2.0 cm, with shortness of breath, mild triconcave sign and double lung breathing sound, no obvious dry and wet rales were heard. The heart sounded powerful. No obvious murmurs were heard in the auscultation area of each valve. The abdomen was soft, the liver and spleen were not swollen, and the umbilical cord was intact. Muscle tone in the extremities was normal, primal reflexes were normal.

Admission diagnosis: Wet lung of newborn?

### 1.2 Laboratory and imaging examination

Blood routine examination on admission: WBC 27.04 × $10^9$/L, N: 80.2%, L: 11.2%, HGB 185 g/L, PLT 235 × $10^9$/L. Blood gas analysis: pH 7:237, $PCO_2$ 32.8 mmHg, $PO_2$ 71.9 mmHg, $HCO_3$act 13.6 mmol/L, BE-12.6, Lac 1.6 mmol/L. Clotting function was generally normal. Chest radiographs showed that a uniform dense shadow was seen in the right middle and upper lung, a little lung tissue shadow was seen in the lower lung, the left lung texture increased, and the right diaphragm and costophrenic angle were not clearly displayed.

Reexamination of chest radiographs on November $15^{th}$ showed low transmittance of the right lung, dense shadow of the right lung, dense shadow of the left upper lung field, increased/blurred texture of the left lower lung, indicating consolidation shadow of the right and left upper lung.

A complete cardiac ultrasound on November $18^{th}$ showed atrial septal defect (4 mm) and stenosis of the left and right branches of the pulmonary artery.

A complete chest CT on November $19^{th}$ showed that the right lung had a faint and fuzzy shadow with unclear boundary, the middle and upper lobe bronchus of the right lung was unclear, the volume of the right lung became smaller, and the mediastinum shifted to the right. The heart shadow was full, and no abnormal enlarged lymph nodes were observed in the hilar mediastinum. There was no obvious thickening of pleura on both sides and no hydrops in both thoracic cavities, indicating the possibility of right lung inflammatory lesions with right lung atelectasis. The right tracheal was found to be excluding possible developmental abnormalities.

## 2. RESULTS

### 2.1 Treatment and outcome

The patient was hospitalized in our department for 9 days and received NCPAP positive pressure ventilation to give oxygen and anti-infection symptomatic treatment. The dyspnea of the child was not significantly relieved, and the heart noise became increasingly obvious. The family of the child requested to be transferred to a higher level hospital for further diagnosis and treatment, and the child was transferred to Ward 4 of the Neonatology Department of the Seventh Medical Center of PLA General Hospital for further treatment on November $22^{nd}$, 2021. After admission, piperacillin and tazobactam were given anti-infection, high-frequency oscillation mode of invasive ventilator, limited fluid volume and other symptomatic treatment. Fiber bronchoscopy showed atelectasis of the right lung and tracheomalacia of the left lung. Echocardiography showed partial anomalous pulmonary vein drainage, atrial septal defect (secondary foramina), inferior vena cava dilatation, pulmonary hypertension, initial coronary artery dilatation, patent ductus arteriosus. CT imaging of pulmonary veins indicated congenital heart disease: partial anomalous pulmonary vein drainage (sub-cardiac), consideration of "machete syndrome," atrial septal defect, consideration of pulmonary hypertension, stenosis of the right pulmonary artery initiation, right lower lung receiving abdominal aorta blood supply, right lung tissue dysplasia, and widening of the inferior vena cava pulmonary veins. Therefore, the comprehensive diagnosis was "machete syndrome." Considering the high surgical risk of the child, conservative symptomatic treatment was recommended. The patient was hospitalized in Ward 4 of the Department of Neonatology, Seventh Medical Center of PLA General Hospital for 22 days and was required to be discharged. The patient died at home 3 months after follow-up.

### 2.2 Literature review

Using "newborn" and "machete syndrome" as keywords, the literatures included in VIP database, CNKI and Wanfang database self-built database were retrieved until November 2021. With "machete syndrome" as the key word, 239 literatures were retrieved from the biomedical literature database "Pubmed web of science database self-built database" in the past 5 years until March 2022.

## 3. DISCUSSION

Machete syndrome is a rare congenital property of abnormal partial or complete pulmonary venous drainage from the right or left lung to the inferior vena cava. With an incidence of approximately 2 in 100,000 live births,[1] the syndrome is commonly associated with right lung dysplasia, pulmonary sequestration, persistent left superior vena cava, and right tilt of the heart. Diagnosis of the syndrome includes: (1) abnormal drainage of all or part of the right pulmonary vein to the inferior vena cava; (2) hypoplasia or malformation of the right pulmonary artery; (3) hypoplasia of the right lung with dextral shift of the heart; (4) abnormal systemic circulation arteries supplying the right lung, originating primarily from the abdominal or lower thoracic aorta. The pathogenesis of the syndrome is still unclear. The abnormal drainage of right lung vein is due to the permanent residual of the pulmonary vein from the connection between the large vein system and the umbilical vitelline vein system in the embryonic period, which leads to the hypoplasia of the right lung and the right displacement of the heart. The latter two are secondary changes. Pulmonary venous malformation may exist alone or in conjunction with other congenital heart diseases. It may also be accompanied by abnormal pulmonary artery development, abnormal pulmonary lobulation, reduced lung volume and branch tracheal diverticulum, bronchial cyst, and diaphragmatic defect. Pulmonary angiography showed high oxygen saturation in the human part of the inferior vena cava and left to right shunt.[2]

Pathology: The ectopically connected pulmonary veins flow back to the right chamber through the inferior vena cava, and the left to right shunt results in increased pulmonary blood, pulmonary hypertension, increased burden on the right heart, and decreased oxygen saturation of the systemic circulation. The patient may present with clinical manifestations of dyspnea, respiratory infection, congestive heart failure, and cyanosis.

Genetics: Both sexes can be affected, female are more than male 2 times, there is a high incidence of family, the disease is autosomal recessive genetic disease.

Machete syndrome is divided into infantile type and child/adult type.[3] The infantile type is diagnosed within 1 year of age, and the symptoms of infantile type usually appear within 2 months after birth, including shortness of breath, repeated pneumonia, developmental delay, and heart failure, etc. If the symptoms are more severe, the prognosis of patients with heart malformations and pulmonary hypertension is poor. The child/adult type is diagnosed after 1 year of age, and the type has late onset of symptoms and may not show any symptoms. The symptoms of children and adults appear late and may not show any symptoms. If the symptoms are severe, it is associated with pulmonary hypertension and lung infection. Due to the different severity of the deformity, the clinical manifestations vary greatly. Most cases belong to the childhood/adult type and are not diagnosed until adolescence or even adulthood. Pulmonary hypertension is often considered to be the cause of poor prognosis. The diagnosis of machete syndrome is usually based on a characteristic chest X-ray and can be confirmed by angiography, however, it is now mainly by transthoracic or transesophageal echocardiography, non-invasive X-ray computed tomography, or magnetic resonance angiography. Fetal echocardiography uses three-dimensional energy Doppler imaging for prenatal diagnosis.[4] Most commonly, patients are asymptomatic in the absence of associated abnormalities and can be treated conservatively. In patients with heart failure, recurrent pneumonia, or pulmonary blood flow ratio greater than 1.5 and high pulmonary hypertension, it is important to re-navigate the right lung vein and repair associated heart defects to avoid progression to right heart failure. Triad of respiratory distress, hypoplasia of the right lung, and right position of the heart should alert clinicians to machete syndrome.

In this case, the child had these triads after birth, however, since the primary hospital did not recognize the disease, the child was clearly diagnosed in the higher hospital. Children with machete syndrome can be treated with surgery in the early neonatal period, but the indications for surgical treatment are still controversial, and the need for surgery is needed should be evaluated according to the severity of the patient's symptoms and pulmonary circulation blood flow.[5] In this case, the disease started in the neonatal period, the operation risk was very high, and the prognosis was poor. The family returned home after careful consideration, and later the patient died 3 months after birth.

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