# NEUROBLASTOMA IN A 10-MONTH INFANT WITH MISDIAGNOSIS OF FAMILY VIOLENCE: A CASE REPORT

Sang Ngoc Nguyen<sup>1</sup>, Lam Vu<sup>1</sup>, and Rang Nguyen<sup>2</sup>

<sup>1</sup>Hai Phong University of Medicine and Pharmacy <sup>2</sup>An Giang General Hospital

April 05, 2024

## Abstract

Neuroblastoma is the most common extracranial solid tumor in children. A 10-month-old boy was presented with neuroblastoma misdiagnosis of family violence bruised and swollen right periorbital region with no obvious proptosis or abnormal eye movements. Neuroblastoma was confirmed with histopathology and bone marrow. After treatment, the symptoms were resolved.

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Nguyen Ngoc Sang<sup>1</sup>, Vu Tung Lam<sup>1</sup>, Nguyen Ngoc Rang<sup>2</sup>

1: Department of Pediatrics, Haiphong University of Medicine and Pharmacy.

Haiphong Children's Hospital.

2: An Giang Central General Hospital.

#### Abstract

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Keywords: neuroblastoma, children, adrenal gland tumor.

Key Clinical Message

Approaching a child with a bruise in the eyes, the first thing to consider is Neuroblastoma which has a differential diagnosis of an accident caused by injuries or "child abuse".

#### 1. Introduction

Neuroblastoma (NBL) is the most common extracranial solid tumor in childhood that derives from the neural crest and clinical symptoms vary depending on location, distal spread, and secreted metabolites of the tumors [1].

NBL is a special disease that occurs almost early in childhood. Neuroblastoma is formed from the abnormal differentiation of germinal cells at embryonic formation and organ formation. This is a disease of the nervous system that can be found throughout the body but usually found in the abdomen or the adrenal medulla. Neuroblastoma is a common renal cancer and is the fourth most common cancer encountered in children,

accounting for 8-10% of all childhood malignancies [2]. Although there are considerable overlaps in symptoms at initial presentation such as abdominal distension, loss of appetite, nausea, or hypertension, children with neuroblastoma usually have a favorable outcome after treatment with chemotherapy and surgery [3]. Described herein was a case of a 10-month-old boy with neuroblastoma presented with increased work of breathing, rhinorrhea, and cough.

#### 2. Case presentation

TD Pham (PTD) was a 10-month-old boy presented with six hours of increased work of breathing, rhinorrhea, and cough lasting for 2 days. Bruising around his right eye was a notable sign at the triage. His grandmother reported that the bruise of the right eye was seen several days after falling from the cot two weeks ago. No medical care was sought for the bruise. He also had a history of irritability and intermittent vomiting over the past two weeks. There was no history of bruising or bleeding, persistent fever, or loss of weight. Since developing increasing work of breathing that morning he had decreased oral intake, though "normal wet nappies". There was no history of allergies and immunizations were up to date.

He was the first child in the family. His father was a drug addict with a history of intravenous drug use (IVDU) during pregnancy. He was born at 33 weeks via Lower segment Cesarean section for a large antepartum hemorrhage. APGARS were 3 and 5 at 1 and 5 minutes respectively. He required intubation and ventilation for 4 hours and followed by 2 days of CPAP.

#### **Physical Examination**

On exam, the infant was in moderate work of breathing with accessory muscle use and tracheal tug. He was afebrile and had the heart rate of 180 beats per minute, respiratory rate of 45 breaths per minute, and a pulse oximetry reading of 98% on room air. Auscultation of the chest revealed air entry bilaterally with scattered inspiratory crepitation. Heart sounds were dual with no additional sounds. The abdomen was soft and movements were symmetrical with normal tone. The right periorbital region was bruised and swollen. There was no obvious proptosis or abnormal eye movements though this was difficult to assess with given his age, so a referral to ophthalmology for further assessment was made. There were no other suggestions for bruising or injury. A mild nappy rash was present. Both body weight and head circumferences were around the 15th percentile.



## Laboratory Investigations

Blood count revealed hemoglobin 10.5 g/dL, White blood cell 14,500/  $\mu$ L (with neutrophil 7,900 /  $\mu$ L), platelet 402,000 /  $\mu$ L. Liver function tests: increased GGT 71 U/L (Ref <50 U/L). Serum UEC, calcium,

phosphate, magnesium, coagulation profile were within normal limits but increased LDH 3825 IU/l (400 – 1100). Urine Catecholamines. Noradren/Creat ratio: 317.4 (16 – 207). A Cranial CT scan revealed a soft tissue mass on the right orbit, measuring 21x28x30mm. An abdominal ultrasound scan revealed a large suprarenal mass and right paraspinal mass. Abdominal CT showed a large right suprarenal mass consistent with neuroblastoma associated with extensive para-aortic lymphadenopathy and liver metastases. Histopathologies of initial aspirates were done with the results: neuroblastoma tumor (stoma poor, poorly differentiated). MYCN amplification detected. Bone marrow confirmed neuroblastoma and MIGB – Metastases to the liver, bone (skull, proximal humerus, distal femur, right tibia, right distal tibia). With all the clues, he was finally diagnosed with stage 4 high-risk neuroblastoma.

#### Management

On admission, a CT head was conducted to evaluate peri-orbital bruising. With the concerning results, an abdominal ultrasound scan and oncology consult were requested. Further investigations confirmed a diagnosis of stage 4, high-risk neuroblastoma was confirmed. An infusaport was inserted and chemotherapy commenced.

He was planned for 5 cycles of induction chemotherapy, with resection of the surgical resection of the primary tumor after cycle 4. Following cycle 2 peripheral stem cell harvest was planned to allow autologous stem cell rescue during the radiotherapy and consolidation phase. He had a prolonged initial inpatient stay due to social concerns. His mother was absent and un-contactable for days at a time, including at times consent was required for treatment. Therefore, in the early days, the treatment and monitoring of the child's progression had faced some difficulties. We had tried to contact the child's grandmother to follow up with the treatment.

#### Progress

He had surgery to remove the tumor and then followed 4 cycles of chemotherapy. Repeat investigations were arranged prior to surgical resection of the primary tumor, showing a 90% volume reduction in the primary tumor, and over 95% reduction in the volume of retroperitoneal and liver metastasis.

PTD underwent open resection of the primary and abdominal metastasis. All macroscopically visible tumors were able to be removed. Histopathology of the primary tumor found necrotic tumor less than 2% viable tumor.

#### 3. Discussion

Neuroblastoma is the second most common abdominal mass in children after Wilms' tumor [4]. It is a neuroendocrine tumor that originates from sympathetic ganglion cells. The most common location for neuroblastoma is on the adrenal glands usually in the adrenal medulla, though it can arise anywhere in the sympathetic nervous system. Neuroblastoma is slightly more common in boys than in girls with a sex ratio of 1.2:15]. Generally, neuroblastoma has no clinical symptoms unless they invade surrounding organs or metastasize. Unique features of these neuroendocrine tumors are the early age of onset, the high frequency of metastatic disease at diagnosis, and the tendency for spontaneous regression of tumors in infancy [6]. Common sites of metastases are lymph nodes, bone, and the liver [7]. Neuroblastoma often presents late, with non-specific signs including an abdominal mass or pain, complications of metastasis to orbits such as proptosis or peri-orbital bruising, unexplained fever and weight loss, anemia, or bone pain [7]. The most important clue for early diagnosis before biopsy or resection is the presence of hypertension. However, currently in our hospital, there is no device to measure blood pressure for young children so that we did not take blood pressure in this case. Hypertension is likely because of the combined effects of tumor secretion of catecholamines, tumor compression of the renal vasculature, and further activation of the renin-angiotensin aldosterone system. Checking urine catecholamine levels (vanillylmandelic acid and homovanillic acid) is very helpful to further correlate these with the possibility of neuroblastoma [8]. CT Scan often shows a large mass extending across the midline, engulfing abdominal vessels and dislocating surrounding structures [4]. In this case, there was a mass of the adrenal gland with the feature consistent with neuroblastoma.

The prognosis of neuroblastoma varies depending on whether the tumor has spread or metastasized (such as

to the liver or bone) [4]. Despite the younger age of onset, which is usually a favorable prognostic indicator, PTD has a high-risk disease, based on his n-MYC status and metastasis to cortical bone [9].

Management of high-risk disease includes induction chemotherapy, local control with surgical resection and radiation, consolidation, and maintenance phases. It has a poor prognosis, with a 5-year survival between 40 - 50% [10].

PTD's case shows the importance of keeping an open mind to a range of differentials, particularly when these include serious diseases. It also illustrates the need to examine the entirety of a child's social situation to ensure there are no limitations to comprehensive care during treatment: PTD was presented to the hospital with six hours of increased work of breathing on a background of two days of rhinorrhea, cough and bruising around his right eye. His father is a drug addict. So, our first impression of his condition could be a case of "child abuse". However, after a careful physical examination, we did not detect any additional bruises on his body. So, we eliminated "child abuse" in our diagnosis.

PTD has now completed his induction chemotherapy. He is living well with his grandmother. Our hospital has to provide financial supports to ensure he is able to attend future appointments and interventions.

## 4. Conclusions

Approaching a child with a bruise in the eyes, the first thing to consider is Neuroblastoma which has a differential diagnosis of an accident caused by injuries or "child abuse". Laboratory tests are required to make a definitive diagnosis and build a treatment plan as soon as possible in order to ensure the best outcome for the patient.

#### **Consent:**

Written informed consent was obtained from the patient's grandmother for publication of these data and the accompanying images. A copy of the written consent is available for review by the Editor of this journal.

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