Open Journal of
Clinical & Medical
Case Reports

Volume 5 (2019)
Issue 2

ISSN: 2379-1039

A rare cause of apnea and tubulopathy

Giovanni Briganti*; Sonia Scaillet

***Giovanni Briganti**
Department of Pathophysiology, Ecole Supérieure de la Santé, Place du Chateau 3, 1014 Lausanne, Switzerland
Phone: 0032 491 25 41 40; Email: giovanni.briganti@hotmail.com

Abstract

We report the case of a three-month old male presenting a polymalformative disorder characterized with dysmorphic facial features including blepharophimosis and associated with apnea, hearing loss and renal tubulopathy. Molecular genetics analyses allowed for the diagnosis of Ohdo-Madokoro-Sonoda syndrome, a poorly described X-linked disease presenting different subtypes. A new variant is hereby described.

Keywords

Ohdo syndrome

Introduction

The early identification of a rare disease involving several malformations and causing systemic consequences through clinical assessment and complementary examination is essential to determine an optimal management in the patient's best interest. This process may be difficult if the disease causing the clinical context is poorly described in the scientific literature or presenting as a novel subtype.

We hereby report the case of a young patient with an extremely rare disorder.

Case Presentation

A three-month old male presented at the emergency department for severe bradycardia (heart rate at 30 bpm) and desaturation at home. Arterial blood analyses showed a severe respiratory acidosis (pH 7.17; pCO_2 86 mmHg), and the patient was hospitalized in the intensive care unit.

The patient was previously followed in a pediatric clinic for nasogastric tube feeding in a context of

an undefined polymalformative syndrome: the main identified malformations were bilateral blepharophimosis, microphthalmia and coloboma in the left eye, microretrognathia, narrow palate, posteriorly rotated ears, pectus excavatum, microphallus, bilateral inguinal hernia, single transverse palmar crease, renal cortical cysts and choroidal plexus cysts. This syndrome was associated with gastroesophageal reflux, hypoglycemia and a renal tubulopathy inducing a hyponatremia.

The child was delivered at 33 weeks of gestational age through a cesarean section (because of a suspicion of chorioamnionitis) from a 33-year old gravida 9 para 3 woman with unilateral hearing loss. His older stepsister presented with language retardation and followed a special education program. The patient's father presented no relevant personal or familial history.

A series of complementary examinations were carried out. The echocardiography showed no sign of patent foramen ovale or left-to-right shunt; EEG and endocrine check-up were normal. An ENT examination showed a transmission hearing loss of 60 dB in the left ear and 55 dB in the right ear, as well as narrow auditive canals bilaterally, rendering the tympanum observation impossible; an inflammation and swelling of the arytenoid cartilages was present, as well as a laryngomalacia. Visual evoked potentials were normal. Auditive potentials revealed badly structured responses at 85 dB; anomalies of wave I supported the presence of additional abnormalities involving internal and middle ear. Somesthetic potentials in the right median nerve showed a very low amplitude. An ophthalmologic examination reported a stenosis of the right tear canal causing a secretion overload; an elevated left eye pressure and dystrophic corneas bilaterally. Guthrie test was negative. A polysomnography revealed a severe obstructive sleep apnea syndrome requiring nasal CPAP.

The patient developed several systemic complications during a 3-month hospitalization period which were corrected before the discharge. Namely, the child developed a multi-factorial respiratory distress syndrome due to hypotonia, a viral pneumonia, and hypersialorrhea; a BIPAP installment was necessary as well as a botulinic toxin injection in the parotid gland (realized under echography surveillance).

The renal tubulopathy presented as a hyponatremia, a hypokalemia and a hypophosphatemia, which were compensated and supplemented.

Neurological complications included hydrocephalus secondary to choroid plexus cysts and cerebral atrophy.

Molecular genetics analyses showed a MED12 genevariant p.Gln2147* (X-linked) and allowed for the diagnosis of Ohdo-Madokoro-Sonoda syndrome.

Discussion

Ohdo-Madokoro-Sonoda syndrome (OMSS) is an extremely rare X-linked disease characterized with dysmorphic facial features such as blepharophimosis, ptosis, epicanthic fold, saddle deformity of the nose, hypoplasia of the maxillary bone and teeth, micrognathia, narrow palate, posteriorly rotated ears, narrow auditive canals. Hear loss is frequently observed. Ophthalmologic abnormalities include microphthalmia,

strabismus. Skeletal abnormalities are reported, such as hip dysplasia or scoliosis. Hypotonia and mental retardation are frequent [1]. Digestive problems such as constipation may also be present. MED12 (mediator complex) was the gene identified as responsible for this syndrome [2]. Recently, variants of this gene were involved in the identification of different types of congenital malformations [3]. OMSS clinical subtypes have also been highlighted in recent cases involving 2 young males [4] and involving hearing loss, like in our case report.

This is, to our knowledge, the first reported case of OMSS associated with renal tubulopathy and renal cortical cysts; it is also the first case in which the variant p.Gln2147* of the MED12 is identified as associated with the disease; this clinical presentation may be considered as a new clinical variant. These findings, which played an important role in the management of complications, may help future clinicians to better describe, manage and anticipate the consequences of this rare syndrome.

Future studies may endeavor to review known cases of OMSS as well as observe the long-term consequences of this rare syndrome.

References

1. Ohdo S, Madokoro H, Sonoda T, Hayakawa K. Mental retardation associated with congenital heart disease, blepharophimosis, blepharoptosis, and hypoplastic teeth. J Med Genet. 1986; 23(3): 242–4.

2. Vulto-van Silfhout AT, de Vries BBA, van Bon BWM, Hoischen A, Ruiterkamp-Versteeg M, Gilissen C, et al. Mutations in MED12 cause X-linked Ohdo syndrome. Am J Hum Genet. 2013; 92(3): 401–6.

3. Narayanan DL, Phadke SR. A novel variant in MED12 gene: Further delineation of phenotype. Am J Med Genet A. 2017; 173(8): 2257–60.

4. Patil SJ, Somashekar PH, Shukla A, Siddaiah S, Bhat V, Girisha KM, et al. Clinical Variability in Familial X-Linked Ohdo Syndrome-Maat-Kievit-Brunner Type with MED12 Mutation. J Pediatr Genet. 2017; 6(3): 198–204.

Manuscript Information: Received: January 06, 2019; Accepted: February 22, 2019; Published: February 28, 2019

Authors Infomation: Giovanni Briganti[1]*; Sonia Scaillet[2]
[1]Department of Pathophysiology, Ecole Supérieure de la Santé, Place du Chateau 3, 1014 Lausanne, Switzerland
[2]Department of Sleep Medicine, Queen Fabiola Children University Hospital, Avenue Jean Joseph Crocq 15, 1020 Bruxelles, Belgium

Citation: Briganti G, Scaillet S. A rare cause of apnea and tubulopathy. Open J Clin Med Case Rep. 2019; 1514.

About the Journal: Open Journal of Clinical and Medical Case Reports is an international, open access, peer reviewed Journal focusing exclusively on case reports covering all areas of clinical & medical sciences.
Visit the journal website at www.jclinmedcasereports.com
For reprints and other information, contact info@jclinmedcasereports.com