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Syndromic Macrosomia Associating Congenital Hydrocephalus, Bilateral Pre-Axial Polydactyly, Dextrocardia And Delayed Development. About A Case.

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Summary
syndromic macrosomia is characterized by excessive fetal growth associated with malformations. It is due to various genetic anomalies; the vital prognosis depends on the types and severity of the malformations or associated factors.
Clinical Case:
This is an average premature newborn of 35 weeks, female, admitted to the neonatology unit for polymalformative syndrome. She was macrosomic, with hydrocephalus, tip shock on the right and 2 bilateral pre-axial supernumerary fingers. Brain CT SCAN revealed supratentorial hydrocephalus with fusion of the ventricles simulating hydranencephaly; Doppler echocardiography and chest x-ray showed dextrocardia. Hence, the diagnosis of syndromic macrosomia.
Keywords: *syndromic macrosomia, hydrocephalus, polydactyly, dextrocardia.*

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I. Introduction

Syndromic macrosomia is a growth abnormality defined by a weight greater than the 90th percentile, plus 2 standard deviations on the reference curve in relation to age or by a weight greater than 4000 grams at birth. It's the consequence of the genetics anomalies [1-3]. Clinically, it's characterized by neonatal macrosomia associated with morphological anomalies (malformations) [4-5]. Worldwide, its prevalence is not well known. To date, a few syndromes have been reported in the literature. These are: Weaver syndrome, Marshall-Smith syndrome, Bardet-Biedl syndrome, Cohen syndrome, Alstrom syndrome, McCune-Albright syndrome or pesudo-hypoparathyroidism, fragile X syndrome, trisomy 2q37, Prader-Willi syndrome and MOMO (Macrosomia Obesity Macrocephaly and Occular Abnomalities) syndrome [6-7]. In Africa and particularly in the Democratic Republic of Congo, data are very rare; a few cases have been described [8]. In Lubumbashi, the frequency of syndromic macrosomia is 4.4%, mortality is 1.5%. The diagnosis is generally made postnatally during the neonatology consultation. However, etiological diagnosis is not possible due to lack of access to molecular biology [9-10]. The interest of our presentation consists in describing a case of atypical syndromic macrosomia associating congenital hydrocephalus, bilateral pre-axial polydactyly and dextrocardia, highlighting the importance of molecular diagnosis in our environment.

II. Case Description

This is an average premature newborn of 35 weeks, female, admitted to the neonatology unit for polymalformative syndrome. She is the second of two siblings, her older brother is in apparent good health. The mother is 36 years old, pauci pare et pauci gesture, with no particular personal or family medical history. The

prenatal consultations were correctly followed, the vaccination schedule respected, 3 ultrasounds were performed, the latest of which revealed macrosomia and hydrocephalus. The father is 52 years old, with no morbid history; the marriage was not consanguineous. The newborn was born from a cesarean section indicated for hydrocephalus, with an APGAR of 8/10/10. The birth weight was 4050 grams (>P97) for a height of 52 cm ((>P97), a head circumference of 56 cm (>P97) and a thoracic circumference of 38 cm. The peak shock was felt on the right and examination of the limbs revealed 2 bilateral pre-axial supernumerary fingers. The diagnosis of syndromic macrosomia was made; cerebral CT SCAN revealed supra-tentorial hydrocephalus with fusion of the ventricles simulating hydranencephaly; cardiac ultrasound Doppler and chest x-ray showed dextrocardia. The evolution during the neonatal period was good. To date, the infant is 17 months old, psychomotor development and bone age are delayed, the Denver score is equivalent to 7 months old. She is awaiting a diversion.

III. Discussion

Syndromic macrosomia is little described worldwide; its frequency is not known. In the literature, some syndromes associated with neonatal macrosomia have been reported, namely:

Wiedemann-Beckwith syndrome (1/13,700 births) due to an anomaly of the 11p15.5 genes, consequence of inactivation of the gene of one of the parents) combines macrosomia, macroglossia, visceromegaly, omphalocele or umbilical hernia and other morphological abnormalities.

Sotos syndrome due to the mutation and deletion of the NSD1 gene located on chromosome 5q35 is characterized by macrosomia, macrocephaly, facial dysmorphism (large bulging forehead), prominent forehead and advanced bone age.

The rarer Weaver syndrome (30 cases described so far) is made up of different facial dysmorphia (micrognatia and filtrum, hyperlaxity with excess skin, hoarse voice, camptodactyly, hypoplastic nail, dental dysplasia).

Marschall-Smith syndrome is made up of statural advancement and bone age, facial dysmorphism (protruding frontal bumps, prominent eyes and blue sclera, antevarsion of the nostrils and micrognatia), skeletal anomalies (abnormal phalanges) and susceptibility to infections respiratory.

Bardet-Biedl syndrome, rare, due to mutation of the BBS1 to 4 genes is characterized by early obesity, retinitis pigmentosa, renal hypoplasia, mental retardation, hypogenitalism.

Prader Willi syndrome, due to a mutation in the proximal region of the long arm of chromosome 15 (15q11-q13) is manifested by neonatal hypotonia, feeding difficulties, facial dysmorphism, hypogenitalism, hyperphagia with overweight, ocular abnormalities and spinal deformity.

Cohen syndrome, rare (200 cases reported), is due to the mutation of the VPS13B gene. it is characterized by facial dysmorphism, microcephaly, truncal obesity, ligamentous hyperlaxity, pigmentary retinopathy and neutropenia.

Alstrom and Mccune Albright syndrome are also considered syndromic macrosomia.

In the Democratic Republic of Congo, 2 phenotypes of syndromic macrosomia have been described, without confirmation of the diagnosis by genetics. Sotos syndrome has been described in Kinshasa and Bekwick-Wiedenann syndrome in Lubumbashi [9-12]. Of all the cases reported in the literature to date, we have not found a case similar to ours, associating neonatal macrosomia, hydrocephalus, bilateral pre-axial polydactyly, dextrocardia and psychomotor devepment delayed. Unfortunately, due to a lack of molecular analysis resources in our environment, a definitive diagnosis could not be made. Hence the question of whether it would be one of the forms of the variants already described or another syndrome in its own right?

IV. Conclusion

Syndromic macrosomia is rarely reported in our environment. We described a case of syndromic macrosomia associating hydrocephalus, dextrocardia and bilateral pre-axial polydactyly and development delayed, an atypical variant not documented in the literature. The lack of access to genetic analyzes did not allow us to document the etiopathogenesis of this association nor to determine the syndrome. Hence the interest in developing molecular biology in our environment.

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