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Case Report

Prenatal diagnosis of binders phenotype: a cross road to decision making

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ABSTRACT

Binder's syndrome is an uncommon congenital condition which develops in the first trimester of pregnancy and has characteristic effects on the facial features. Those effects are: arhinoid face, intermaxillary hypoplasia (associated with malocclusion), abnormal position of the nasal bones, nasal mucosa atrophy, anterior nasal spine agenesis and (in most cases) a lack of frontal sinuses. Other deformities, as well as mental retardation, are also possible. Due to the rarity of the disease, there are no treatment trials for these patients. Treatments reported in the medical literature are part of single case reports or small case series of patients. Here is a case report of a 23-year-old who presented to OPD at 26 weeks with features of binders' phenotype. This woman delivered an infant of 2.7 kg without other abnormalities and no respiratory distress postnatally. Sonographic evaluation during pregnancy depicts proper differential diagnosis and therapeutic strategy. Multidisciplinary approach is mandatory in order to establish meticulous treatment. Further studies must be conducted, achieving this ultimate scope. Even so, the parents should be counselled that Binder syndrome is a tentative diagnosis prenatally, and that not all genetic syndromes can be prenatally excluded.

Keywords: Binders phenotype, Binders syndrome, Intermaxillary hypoplasia, Congenital condition

INTRODUCTION

The Binder phenotype is defined by midface hypoplasia, underdeveloped frontal sinuses, hypoplastic and abnormally positioned nasal bones, nostrils appearing moon or comma-shaped and prognathism. It is heterogeneous in etiology and not fully understood. Multiple causative factors are described. The physician's dilemma is of diagnosing the fetal abnormalities on antenatal ultrasound for which there is no confirmatory testing. There are management and ethical problems regarding the diagnosis, further investigations and confirmation of the diagnosis. The Binder phenotype is a clinical diagnosis with multiple differential diagnoses. It carries an uncertain and unpredictable course and prognosis which might be difficult for both clinician and parents to predict and comprehend. Genetic counselling has to be variable depending upon the diagnosis.[1]

CASE REPORT

A 23 year old G3P1D1A1 presented to antenatal OPD at a gestation of 26 weeks+1day whose first pregnancy resulted in stillbirth at 26 weeks of gestation and her second pregnancy was terminated at 22 weeks of gestation and now she came for second opinion as her NT scan showed hypoplastic nasal bone and flat facies, Negative double markers, TIFFA showing features of binders phenotype i.e., nasal and facial hypoplasia with frontonasal facial angle of 1530 along with echogenic intracardiac foci in left ventricle following which she was adviced the couple were counselled about the tentative diagnosis of Binder syndrome, its potential implications

and the limitations of prenatal diagnosis. Amniocentesis with array comparative hybridization (aCGH) was performed, indicating a normal female karyotype. At follow-up scans, no additional abnormalities were detected and the biometry of the long bones remained within normal limits. The course of pregnancy was uneventful, and an infant of 2750 g was born at 39+2 gestational weeks by normal vaginal delivery. At two months of life, the infant was doing well, and the only complaint at the time was tedious breathing and episodes of postprandial cough.

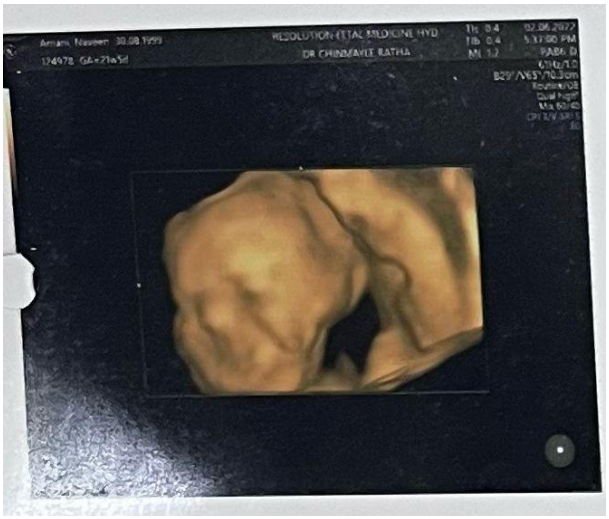

Figure 1: 3D Ultrasonographic examination of fetus showing nasal hypoplasia at 24 weeks.

Figure 2: Postnatal image of the baby.

DISCUSSION

There are only few published cases with prenatal diagnosis of Binder syndrome. The difficulties faced by examiners and parents after the identification of a Binder phenotype at the fetal anomaly scan mostly focus on three issues, i.e., ascertainment of the diagnosis, obstetric management and prognosis.

Binder syndrome was first diagnosed in 2000 and encompasses verticalisation of nasal bone, short columella, flattened nose and frequently associated with prognathism of the mandible. The length of the nasal bone is usually normal. The facial anomalies can be isolated or associated with other features, commonly skeletal abnormalities such as stippled epiphysis, rhizomelic shortening and cervical defects. Strabismus, hearing deficiency, microdontia of the upper incisors can appear later in life. Diagnosis is usually made on ultrasound scan, sagittal section.

A careful evaluation of foetal anatomy should be undertaken, especially skeletal and heart assessment three dimensional images help evaluate the face and diagnose binder syndrome. MRI would bring additional information in cases of skeletal defects. Cases of Binder phenotype with prenatal suspicion of vertebral stenosis or cervicothoracic kyphosis on the antenatal US and Magnetic resonance imaging (MRI), have been described as part of chondrodysplasia punctata (CP) which is the principal differential diagnosis. Radiographic findings of cartilaginous stippling due to due to calcium build-up during endochondral bone development are the central clue for chondrodysplasia punctata diagnosis. There are several ways of CP inheritance in an autosomal dominant manner, X linked dominant, and X linked recessive. Imaging diagnosis of Binder syndrome should led to a detailed review of antenatal and pre-pregnancy history, conditions associated with decreased levels of Vitamin K, such as hepatic disease, alcoholism, hyperemesis gravidarum. More than 50 conditions associated with low flat nasal bones have been described although, most of them have other associated anomalies. Among genetic conditions that have a flat profile apart chondrodysplasia punctate are Wolff-Hirschhorn syndrome (Greek warrior helmet), Keutel and Robinow syndrome craniosynostosis and fetal warfarin disorder.[3]

Introduction of non-invasive prenatal testing (NIPT) in clinical practice has widened screening possibilities advocating that cell-free DNA (cf DNA), in many cases, is a replacement of invasive techniques. Perinatal respiratory problems have been reported in foetuses with spine and cervical abnormalities. An article published by Blumfielled at all reports three cases of babies with respiratory distress post-delivery, which should not be surprising as there are spinal, neck and head abnormalities. Nevertheless, when isolated Binder syndrome has an excellent prognosis, with teenagers seeking medical assistance rather for cosmetic reasons.

Prognosis

Hearing loss has been reported in 5% of Binder syndrome cases. The syndrome's unique facial defects can be surgically corrected using a multi-stage multi-disciplinary approach which has a better prognosis if started early in life. Postponing surgical treatment results in a missed opportunity of normal-like facial growth and prolongation

of the psychological burden, imposed by a negative self-image. Mental retardation, although reported in limited cases, it is not considered a significant feature of the syndrome.[4]

It is important that antenatally diagnosed Binder Phenotype cannot definitely exclude more serious conditions, potentially involving neurodevelopmental delay. As noted before, out of eight cases of Binder phenotype antenatally diagnosed by Levaillant et al, two (25%) had an isolated Binder phenotype, while the remaining cases were related conditions sharing a similar phenotype.[3]

Obstetric and perinatal care

No specific modification of the obstetric care is required. Recently, Blumenfeld et al reported three cases of antenatally diagnosed Binder phenotype associated to chondrodysplasia punctata, complicated by respiratory distress during the immediate post-natal period, attributed to obstruction of the narrowed upper airway. Therefore, care should be taken for preparation for neonatal resuscitation in such cases.[4]

Differential diagnosis

The differential diagnosis of Binder syndrome includes other facial disorders, most of which are usually associated with genetic disorders. Binder syndrome should be suspected when prenatal ultrasound identifies a low flat nasal bridge. Chondrodysplasia punctata is also associated with scoliosis and asymmetrical shortening of the limbs. Robinow syndrome is associated with short forearms, clinodactyly, and macrocephaly. Aarskog syndrome is associated with brachycephaly and clinodactyly of the 5th finger. Crouzon syndrome is associated with craniosynostosis and short occipital-frontal diameter. Apert syndrome is characterized by irregular craniosynostosis, short occipital-frontal diameter, flat occiput, ventriculomegaly, and syndactyly. Rudiger syndrome presents with short digits and talipes. Keutel syndrome is characterized by calcification and/or ossification of the cartilage in the external ears, nose, larynx, epiglottis, thyroid, trachea, ala nasi, and ribs; this syndrome also presents with peripheral pulmonary stenosis, sensory hearing loss, and borderline-to-mild mental retardation.

CONCLUSION

Binder Syndrome represents a controversial entity with variety of phenotypic abnormalities. Due to the rarity of the disease, there are no treatment trials for these patients. Treatments reported in the medical literature are part of single case reports or small case series of patients.

Sonographic evaluation during pregnancy depicts proper differential diagnosis and therapeutic strategy. Multidisciplinary approach is mandatory in order to establish meticulous treatment. Further studies must be conducted, achieving this ultimate scope. Even so, the parents should be counselled that Binder syndrome is a tentative diagnosis prenatally, and that not all genetic syndromes can be prenatally excluded. Faced with this uncertainty, many parents would opt for pregnancy termination; among a total of 16 published cases with prenatal diagnosis, 6 (38%) chose for a termination.

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