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### Case Study

## MIDLINE FUSION DEFECT

Kulkarni Meghana<sup>1\*</sup>, Aher Gautam<sup>2</sup>

<sup>1</sup>Resident, Dept of Obstetrics & Gynecology; Padmashree Dr. Vithalrao Vikhe Patil Medical College & Hospital, Vilad Ghat, Ahmednagar – 414111

<sup>2</sup>Professor, Dept of Obstetrics & Gynecology; Padmashree Dr. Vithalrao Vikhe Patil Medical College & Hospital, Vilad Ghat, Ahmednagar – 414111

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\*Corresponding Author: **Dr. Meghana Kulkarni**

Dept of Obstetrics & Gynecology; Padmashree Dr. Vithalrao Vikhe Patil Medical College & Hospital, Near Govt. Milk Dairy, Vilad Ghat, Ahmednagar – 414111

E-mail: [meghana1787@yahoo.co.in](mailto:meghana1787@yahoo.co.in) Mobile No.: 8237649416

### ABSTRACT

Midline fusion defect is failure to fuse two embryonic halves. Primary palate forms during 4<sup>th</sup> – 7<sup>th</sup> week of gestation when two maxillary & medial nasal swellings fuse. Failure of this fusion leads to midline facial fusion defects. Gastroschisis is evidenced by a hole in the baby's abdominal wall that the intestines stick out of. This condition is caused by a defect on one side of the umbilical cord. It looks similar to an omphalocele, except that it is usually smaller and organs are not covered by a membrane. Gastroschisis exposes the baby's intestines to amniotic fluid before birth, which increases the risk for complications including bowel dilation, decreased fetal growth and amniotic fluid volume, preterm delivery, and, rarely, fetal death. The condition is usually diagnosed through an ultrasound before birth. Our case is rarest form of Gastroschisis associated with extra abdominal midline fusion like cleft lip, cleft palate with temporal bone deformity.

**Keywords:** Midline fusion defect, Gastroschisis, Extra abdominal fusion defect, Cleft lip, Cleft palate.

### INTRODUCTION

Described in the literature as early as the first century AD, today these anomalies are frequently detected prenatally due to routine maternal serum screening and fetal ultrasound. Gastroschisis represents a herniation of abdominal contents through a paramedian full-thickness abdominal fusion defect. The abdominal herniation is usually to the right of the umbilical cord. No genetic association exists. A gastroschisis usually contains small bowel and has no surrounding membrane. The herniated bowel is not rotated and is devoid of secondary fixation to the posterior abdominal wall.<sup>1</sup> Because the herniated bowel is bathed by amniotic fluid, both maternal serum and amniotic fluid alpha-fetoprotein (AFP) levels are elevated, more so than in exomphalos. Thus, gastroschisis is found incidentally or because of an elevated maternal AFP level, a finding in 77-100% of cases. Rarely, polyhydramnios may prompt an antenatal sonographic examination. Fetal growth restriction is a frequent association. Oligohydramnios is rare. Chromosomal anomalies are not associated with gastroschisis, and familial occurrence is exceptionally rare. In very rare instance Gastroschisis is associated with extra-abdominal midline defect like cleft lip & cleft palate. Primary palate forms during 4<sup>th</sup> – 7<sup>th</sup> week of gestation when two maxillary & medial nasal swellings fuse. Failure of this fusion leads to midline facial fusion defects. Our case is one of such rarest presentation.

### CASE SUMMARY

We present a case of gastroschisis associated with cleft lip & cleft palate.

A 23-year-old primigravida was referred to our hospital after routine dating ultrasound that revealed anomaly. The patient had no antenatal care prior to this time and was 32 weeks by dates. The family history was negative for congenital anomalies or genetic abnormalities and the patient denied exposure to drugs or toxins.

Our ultrasound examination revealed a single fetus with 30 weeks gestational age with cleft lip & cleft palate associated with gastroschisis. The temporal bone defect was large. We didn't notice any signs of obstruction or peritonitis.

The pregnancy was terminated in view of congenital anomaly as the patient was not willing for continuation. Infant was female.

**The particularity of our case is association with cleft lip & cleft palate.**

### DISCUSSION

Gastroschisis represents a congenital defect characterized by a defect in the anterior abdominal wall through which the abdominal contents freely protrude. There is no overlying sac and the size of the defect is usually less than 4 centimetres (1.6 in). The abdominal wall defect is located at the junction

of the umbilicus and normal skin, and is almost always to the right of the umbilicus<sup>1</sup>.

Widespread use of antenatal ultrasound examination and maternal serum alpha-fetoprotein (MSAFP) screening has made the detection of gastroschisis possible in the second trimester of pregnancy<sup>2</sup>.

**Omphalocele** is another congenital birth defect, but it involves the umbilical cord itself, and the organs remain enclosed in visceral peritoneum. With omphalocele the defect is usually much larger than in gastroschisis.

#### CAUSES

High-risk pregnancies such as those complicated by infection, young maternal age, smoking, drug abuse, or anything that contributes to low birth weight can increase the incidence of gastroschisis, which is more frequent in newborns who are small for gestational age. Whether the intrauterine growth retardation can facilitate the apparition of gastroschisis or the abdominal wall defect impairs fetal growth is not clear yet. Numerous clinical studies have linked aspirin, a U.S. Food and Drug Administration (FDA) pregnancy category D drug, as an increased risk factor<sup>3,4,5</sup>, and according to large scale study by the California Department of Public Health aspirin quadruples the risk of Gastroschisis<sup>6</sup>.

A change in paternity (childbearing with different fathers) has been implicated as a risk factor in a recent study, suggesting that the immune system of the mother may play a role in the development of gastroschisis<sup>7</sup>.

#### GENETICS

Gastroschisis as a stand-alone congenital defect, like many birth defects when isolated, exhibits multifactorial determination with a 2-3% recurrence risk for subsequent pregnancies. Rarely, gastroschisis has clustered in families and exhibits autosomal recessive or dominant inheritance.<sup>8</sup>

Genetic counseling and further genetic testing, such as amniocentesis, may be offered during the pregnancy, as this and other abdominal wall defects are associated with genetic disorders. If there are no additional genetic problems or birth defects, surgery soon after birth can often repair the opening.

#### PATHOPHYSIOLOGY

At least six hypotheses have been proposed:

1. Failure of mesoderm to form in the body wall<sup>9</sup>
2. Rupture of the amnion around the umbilical ring with subsequent herniation of bowel<sup>10</sup>
3. Abnormal involution of the right umbilical vein leading to weakening of the body wall and gut herniation<sup>11</sup>
4. Disruption of the right vitelline (yolk sac) artery with subsequent body wall damage and gut herniation<sup>12</sup>
5. Abnormal folding of the body wall results in a ventral body wall defect through which the gut herniates<sup>13</sup>
6. Failure to incorporate the yolk sac and related vitelline structures into the yolk sac<sup>14</sup>

The first hypothesis does not explain why the mesoderm defect would occur in such a specific small area. The second hypothesis does not explain the low percentage of associated abnormality compared with omphalocele. The third hypothesis was criticized due to no vascular supplement of anterior abdominal wall by umbilical vein. The fourth hypothesis was commonly accepted, but it was later shown that the right

vitelline artery (right omphalomesenteric artery) did not supply the anterior abdominal wall in this area.<sup>15</sup> More evidence is needed to support the fifth hypothesis<sup>13</sup>.

#### EMBRYOLOGY

During the fourth week of development, the lateral body folds move ventrally and fuse in the midline to form the anterior body wall. Incomplete fusion results in a defect that allows abdominal viscera to protrude through the abdominal wall. The bowel typically herniates through the rectus muscle, lying to the right of the umbilicus.

#### TREATMENT

Gastroschisis is a surgical emergency<sup>16</sup>. Patients frequently require more than one surgery; only about 10 percent of cases can be closed in a single surgery<sup>16</sup>.

The general procedure for gastroschisis is to simply tuck the protruding organs back into the opening and apply a belly band pressure until the wound heals itself. New advances have been pioneered in repairing the protruding bowel by placing a protective "silo" around the intestine outside the abdomen, then slowly pressuring the herniated intestine into the abdominal cavity. This new procedure allows for the bowel to return to its intended shape and location without further traumatizing the infant's viscera with undue internal pressure. The main cause for lengthy recovery periods in patients is the time taken for the infants' bowel function to return to normal.

#### PROGNOSIS

Current advances in surgical techniques and intensive care management for neonates have increased the survival rate to 90%,<sup>17,18</sup> in adequate settings. The possibility of prenatal diagnosis either through echosonogram or any other method available allows the mother to be referred to an adequate center where a caesarean section or induced natural birth can be performed before term (as natural birth is recommended and just as safe as with a normal baby), preferably within 2 weeks of term, and allow the immediate surgery to be performed on the newborn.

The morbidity is closely related to the presence of other malformations and complications of the wound or the intestine.

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Figure 1: Cleft lip and Clef Plate



Figure 2: Herniation of abdominal content through anterior abdominal wall defect



Figure 3: Right lateral view showing temporal bone fusion defect

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