University Journal of Pre and Para Clinical Sciences

ISSN 2455–2879 2020, Vol.6(3)

SACRAL DYSGENESIS WITH ANO-RECTAL MALFORMATIONS

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Abstract : Sacral dysgenesis manifests as dysplasia, hypoplasia or absence of sacral vertebrae. Sacrum is derived from medio ventral portions of somites 30-35, which forms a series of ossification centres that are initially visualized as radiographic opacities by 14 wks of gestation. Sacral dysgenesis usually occurs in association with other anomalies like VACTRL Association and Asymmelia. Sacral dysgenesis with ano-rectal malformations was observed in one spontaneously aborted fetus of 20 wks. Such type of anomalies is very rare and is clinically important for surgeons and pediatricians.

Keyword : Sacral dysgenesis, Ano-rectal malformation.

INTRODUCTION:

A wide range of congenital ano-rectal anomalies are quite common and frequently reported in literature. Sacral agenesis is explained as the absence of lower coccygeal segments to complete aplasia of the vertebrae below T12 segment. Sacral dysgenesis with ano-rectal malformations is explained as the related conditions (1). The vertebral column is derived from 44 somites of the paraxial mesoderm. These mesodermal somatomeres occur in conjunction with primary neurulation down to the 30th somite. The 30th somite corresponds to S1-S2 junction. From S2 segment till the coccyx is derived from the somites 31 to 44 and arises from the caudal eminence (tail bud) during secondary neurulation and the retrogressive differentiation that follows. The ossification of vertebral column proceeds in a cranio-caudal sequence. Then ossification proceeds every 2 to 3wks.So S2 is ossified by 22wks (2).The endodermal cloaca or post allantoic part forms the rectum and upper part of anal canal. Uro- Rectal septum divides the endodermal cloaca into ventral part, the primitive urogenital sinus and a dorsal part, the primitive rectum. The urorectal septum extends towards the cloacal membrane. A gap, cloacal duct appears between the caudal free edge of the urorectal septum and the cloacal membrane. At 7th week the cloacal duct is closed and the urorectal septum fuses with the cloacal membrane. So the cloacal membrane is divided into urogenital membrane in front and the anal membrane behind. The endodermal cloaca behind the perineal body presents a surface depression, the proctodeum or anal pit. The anal canal is developed from 2 sources. Upper part from endodermal cloaca and the lower part from the ectodermal proctodeum (2). Malformation of sacral vertebrae occurs very rarely. Maternal diabetes, genetic predisposition and vascular hypoperfusion have been suggested as causative factors (3). Sacral dysgenesis with ano-rectal malformations is reported to occur 1 in 5000 live births (4). Here I am reporting a case of sacral dysgenesis with ano-rectal malformations in a 20wks aborted fetus.

MATERIALS AND METHODS:

20wks aborted fetus was procured from the Obstretics and Gynaecology department and was preserved in 10% formalin. The fetus was examined and dissected for any external, skeletal and internal organ abnormalities.

OBSERVATIONS:

The following abnormalities were found in a 20wks aborted fetus. The fetus was observed to have sacral dysgenesis (Fig 1 and Fig 4), absence of anal canal (Fig 5) and the rectum was found to be distended (Fig 3).

DISCUSSION:

It has been explained that sacral agenesis is absence of the terminal segments of the vertebral column, ranging from absence of lower coccygeal segments to complete aplasia of the vertebrae below T12 segment. Related conditions are sacral dysgenesis in which part of sacrum is absent and caudal regression (Dysplasia, Deficiency and Dysgenesis), in which ano-rectal malformations may be present(1). Coinciding with the above report, sacral dysgenesis was observed in the present case reported here also . Standring(2008) explains that ossification of vertebral column proceeds in a cranio caudal sequence. After 16wks it spreads to L5.Then ossification proceeds every 2 to 3 wks. So S2 is ossified by 22wks.Very rarely sacral vertebral malformations develop, with an association of maternal history of diabetes mellitus(2). In contrary to the above report, the present case reported here was observed to have a wide developmental defect in the sacrum in a 20wks aborted fetus as found in Fig 4, which is unlikely to be ossified by 22wks and correlating with the other associated anomalies like distended sacrum and absence of anal opening this case was diagnosed as a case of Sacral dysgenesis . Aslan et al (2001) has reported a case of prenatal diagnosis of caudal regression syndrome where a 16yr old primigravida at 22wks of gestation presented with a singleton fetus with normal amniotic fluid in USG examination. A sudden termination of spine at lumbar level and fixed lower extremeties with club feet with flexion contractures of the lower extremeties

with extension popliteal webbing in frog like position was noted and reported as caudal regression syndrome. This developmental abnormality occurs at the midposterior axial mesoderm and the lesion is said to originate before the 4th week of gestation (3). Rong et al (2001) has done genetic studies in mice and has reported the reason for human anorectal malformations as defect in Shh signaling and has found out multiple coding region of HLXB9 in patients with sacral agenesis (4).Nour et al (1989) has reported that the Incidence of ano-rectal anomalies is 1 in 5000 live births. This congenital anomaly is usually associated with bony defects in the sacrum. The bony defect varies from complete to partial sacral agenesis to spina bifida occulta or meningomyelocoele. They have reported low ano-rectal anomalies with partial sacral agenesis in 2 of 6 brothers(5). In this case rectum was found to be distended and anal opening was absent along with Sacral dysgenesis . Wang et al (1999) have reported the sacral dysgenesis as a sporadic event. They have identified 2 families with underlying chromosome 7q deletion causing sacral dysgenesis. All affected individuals had developmental delay and microcephaly. They also explained that Curarrino syndrome represents only 5% of symptomatic patients with ano-rectal malformations. But the involvement of urogenital system and lower GIT is very similar to that observed with sporadically occurring sacral dysgenesis(6). Sacral dysgenesis observed in the present case was coinciding with the above report, but it was observed in a 20wks aborted fetus . Sacral Agenesis with anorectal malformations has been frequently reported in literature as caudal regression syndrome or as part of currarino syndrome. But in the case reported here, sacrum was not fully formed and the rectum was found to be distended and anal opening was found to be absent. Lynch et al (2000) has reported sacral agenesis as autosomal dominant trait, which shows partial agenesis of sacrum involving S2, S3 vertebrae. He has also reported HLXB9 as a major gene causing curarrino syndrome which has associated features like anorectal malformations, a presacral mass and urogenital malformations (7). The etiology of this condition is not clearly reported in the literature. Chromosomal studies reported in journals have explained the cause as the terminal deletion of chromosome 7q or defect in sonic hedgehog signaling or HLXB9 as a major gene causing curarrino syndrome.

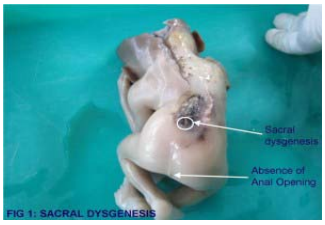

FIG 1: SACRAL DYSGENESIS

FIG 2: SACRAL DYSGENESIS ALONG WITH SPINAL CORD

FIG 3: DISTENDED RECTUM

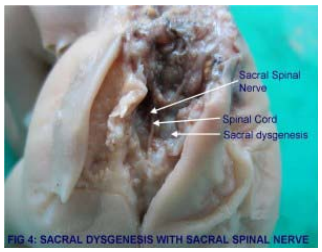

FIG 4: SACRAL DYSGENESIS WITH SACRAL SPINAL NERVE

FIG 5: ABSENCE OF ANAL OPENING

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University Journal of Pre and Para Clinical Sciences