

CLINICAL IMAGE

Perineal Groove with Neural Tube Defect: A Rarer Finding

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A 5-day-old female baby born at 34 weeks of gestation, birth weight 2.6 kg, was brought to us with complain of lower back midline swelling since birth. On examination, the baby had lumbosacral myelomeningocele (MMC) and a mucosa-lined perineal groove (Figure 1). Anal index calculated was 0.34 and no neurogenic bowel or bladder component. The baby underwent excision and repair of MMC and perineal groove was left as such to epithelize.

The perineal groove is a very rare type of anorectal malformation with unknown incidence. It was first described by Stephens in 1968 [1]. The etiopathogenesis of this rare anomaly remains largely illusive. Various theories postulated are the developmental defect of urorectal septum [2]; failure of fusion of medial genital folds in midline [1]. The histologic examinations of resected specimens have revealed simple columnar or stratified columnar or cuboidal epithelium of a rectal type mucosa to a non-keratinized stratified squamous epithelium [1,3]. This congenital anomaly has tendency of self-epithelialization, usually by 1–2 years of age, and surgery is reserved mainly for cosmetic grounds.

The perineal groove may be misdiagnosed as inflammation, dermatitis, injury, etc. It is imperative to recognize this anomaly at birth for appropriate parental counseling and to prevent untoward surgery or medical management.



Figure 1: LS myelomeningocele with perineal groove showing midline mucosal defect connecting anus to vestibule

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