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VAGINAL ATRESIA- ONE AMONGST THE MANIFESTATION OF RARE “CHARGE” SYNDROME

CHARGE syndrome is a rare genetic disorder caused due to mutation in CHD7 gene- an ATP dependant chromatin remodeler. CHARGE itself abbreviates its manifestation, C- Coloboma/ Microphthalmia, H- Heart defects, A-Atresia Choana, R- Retardation of growth, G- Genital abnormalities, E- Ear abnormalities. A 15yr old girl with primary amenorrhoea presented to OBG OPD in April 2019 with complaints of cyclical lower abdominal pain since 7 months. On GPE & systemic examination- 1. She had left microphthalmia with vision of only perception of light. 2. Characteristic LOP ear was noted with h/o reduced hearing since childhood. 3. Secondary sexual characters, however revealed Tanner Stage 3- normal for her age. 4. CVS revealed Atrial septal defect. After proper consenting from the guardians, local examination revealed Vaginal atresia. After CTVS consultation, she underwent ASD correction in May 2019. Further she was subjected to imaging studies like USG & MRI abdomen and pelvis, which revealed a normal uterus and adnexa with hematometra and cervix appearing quite small in size, also noticing a relatively small left kidney with mild pelvicalceal fullness. After all initial investigations, confirmation of diagnosis and pre anesthetic clearance, she was planned for Vaginoplasty after 3 months. On 7-8-19, she underwent Mc Indoe’s vaginoplasty with hematometra being drained by hysterotomy & upper part of neovagina was anastomosed with cervical opening, a split skin graft harvested from the left thigh posterior aspect was used to create a neovagina. I want to present this case due to its rarity and successful management.

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