Abstract: 19 years old girl admitted with co not attained menarche. On examination secondary sexual characters are present appropriate to her age, general examination including height, blood pressure, CVS, RS, CNS, external genitalia normal. Blood investigations including thyroid profile, FSH, LH, prolactin, testosterone normal. Ultrasound, MRI showed hypo plastic uterus, normal ovaries. Karyotyping showed 46XX. Examination under anaesthesia revealed blind vagina of 2cm.on per rectal examination no distinct uterine structure felt. Diagnostic laparoscopy showed rudimentary uterine bud, bilateral normal ovaries. The patient was advised perineal dilation (Frank technique).

Keyword: Amenorrhea, Mullerian agenesis, Utero vaginal atresia, perineal dilation

Case Summary: 19 year old girl presented with complaints of not attained menarche. NO present h/o cyclical lower abdominal pain, social psychological problem, headache, anosmia, auditory dysfunction, visual disturbances, milk secretion, excessive eating followed by vomiting, malnutrition, excessive weight gain or loss, vigorous exercises, thyroid disorder, brain irradiation, surgery, head injury, drug intake. Patient born of non consanguineous marriage, birth weight 2.8kg, no history of prolonged labour, milestones normal, school performances normal, consumes veg and non veg diet, bowel and bladder habits normal, sleep pattern normal. Mother attained menarche at 14 years of age. Her younger sister attained menarche at 13 years of age. No family history of premature ovarian failure .No past history of long term drug intake, tuberculosis, Epilepsy, Heart disease, Thyroid surgery

ON EXAMINATION: Moderately built, moderately nourished, height 158cm,weight 45kg,BMI 18,arm span 148cm,breast and pubic hairs tanner stage 5,axillary hair present, no masculine features. Breast and thyroid normal, no features suggestive of turner syndrome, vitals normal ,CNS ,RS ,CVS ,vision ,olfactory auditory system normal, per abdomen soft and no organomegaly, external genitalia normal.

INVESTIGATIONS: Blood investigations including hormonal assays normal. Karyotyping 46XX. MRI showed rudimentary uterus, bilateral ovaries normal, diagnostic laparoscopy revealed rudimentary uterine bud, bilateral normal appearing ovaries.

LAPROSCOPIC PICTURES

Mayer Rokitansky Kuster Hauser (MRKH) syndrome -CASE REPORT
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DISCUSSION:
Mayer Rokitansky Kuster Hauser (MRKH) syndrome (Mullerian agenesis) is a disorder caused by incomplete development or complete absence of mullerian duct. This condition causes the vagina and uterus to be underdeveloped or absent (MRKH Type 1). In 30-40% cases of women with MRKH syndrome may also have abnormalities in other systems. The kidneys may be abnormally formed or positioned, or one kidney may fail to develop (unilateral renal agenesis). They also commonly develop skeletal abnormalities, particularly of vertebrae, hearing loss or cardiac abnormalities (MRKH Type 2). Incidence is approximately 1 in 4,500. The cause of MRKH syndrome is unknown, although it probably results from a combination of genetic and environmental factors. Wt1, Pax2, Cfr and Hox genes may be associated with MRKH syndrome. In some families, the condition appears to have an autosomal dominant pattern of inheritance. Often, the first noticeable sign of MRKH syndrome is that menstruation does not begin by age 16 (primary amenorrhea). Women have chromosomal pattern 46XX and normally functioning ovaries. They also have normal female external genitalia, normal breast and pubic hair development and normal hormonal assays.

MANAGEMENT:
Diagnosis
Ø Chromosomal analysis to exclude karyotypic abnormalities of the X chromosome and androgen insensitivity syndrome (AIS); individuals with complete AIS have female external genitalia but a 46,XY karyotype. Circulating levels of luteinizing hormone (LH) and follicle-stimulating hormone (FSH), which are normal in MRKH syndrome, indicating appropriate ovarian function. Testosterone levels can be assayed and are in the normal female range
Ø Imaging modalities used for MRKH syndrome include the following: Ultrasonography, MRI, Laparoscopy, Pyelography

Treatment
The goal of treatment is to provide the patient with an unscarred vagina that allows sexual functioning

NON SURGICAL METHOD
Frank technique or perineal dilation

SURGICAL METHODS
McIndoe technique, Williams’s vaginoplasty, Rotational flap procedures, Intestinal neovagina, and Vecchietti technique. Excision of uterine anlage can also prevent endometriosis and resultant ovarian function impairment. Although women with this condition are usually unable to carry a pregnancy, they may be able to have children through assisted reproduction.

REFERENCES: