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The Medical Bulletin

In Syndrome

Mayer-Rokitansky-Kuster-Hauser Syndrome Synonyms :

- CAUV (Congenital Absence of the Uterus and Vagina)
- MA (Müllerian Aplasia)
- GRES (Genital Renal Ear Syndrome)

Definition :

- The MRKH syndrome is characterized by congenital aplasia of the uterus and the upper part (2/3) of the vagina in women with normal development of secondary sexual characteristics and a normal 46, XX karyotype.

Etiology :

- It's remained quite unclear until now.
- Aberrant expression of anti-Müllerian hormone (AMH) or its receptor, both involved in Müllerian duct regression was hypothesized as a cause of MRKH syndrome.
- Also a loss-of-function mutation in the WNT4 gene.

Epidemiology : 1 in 4500 female births.

Types :

- Type I (isolated) or Rokitansky sequence
- Type II or MURCS association (Mullerian duct aplasia, Renal dysplasia and Cervical Somite anomalies)

Clinical features : Principle features of MRKH

Syndrome :

- Primary amenorrhea
- Complete uterus aplasia in the presence of two rudimentary horns linked by a peritoneal fold and normal Fallopian tubes correspond to isolated or MRKH type I syndrome.
- Type II MRKH - uterine symmetric or asymmetric hypoplasia, accompanied by aplasia of one of the two horns or by a size difference between the two horn rudiments, coupled with tubar malformations such as hypoplasia or aplasia of one or the two tubes.
- Associated malformations in MRKH syndrome type II (MURCS association)

- Associated upper urinary tract malformations
  - ❖ Unilateral renal agenesis (23– 28%)
  - ❖ Ectopia of one or both kidneys (17%)
  - ❖ Renal hypoplasia (4%)
  - ❖ Horseshoe kidney
  - ❖ Hydronephrosis
- Associated skeletal abnormalities
  - ❖ Spinal anomalies (30 to 40%) - scoliosis (20%), isolated vertebral anomalies (asymmetric, fused or wedged vertebrae), Klippel-Feil association (fusion of at least two cervical segments, short neck, low hair line, restriction of neck motion) and/or Sprengel's deformity, rib malformation or agenesis, and spina bifida.
  - ❖ Anomalies of face and limb extremities - brachymesophalangy, ectrodactyly, duplicated thumb, absent radius, atrio-digital dysplasia (Holt-Oram like syndrome) and facial asymmetry.
- Associated hearing impairment - Auditory defects or deafness 10 to 25%.
- Associated heart malformations -less common.

Diagnosis :

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➢ Transabdominal ultrasonography reveals an absence of the uterine structure between the bladder and the rectum.

➢ Magnetic resonance imaging allows an accurate evaluation of the uterine aplasia, as well as a clear visualization of the rudimentary horns and ovaries. The uterine aplasia is best characterized on sagittal images, while vaginal aplasia is best evidenced on transverse images. Moreover, MRI can be used at the same time to search for associated renal and skeletal malformations.

➢ Celioscopy defines the precise anatomical location and abnormalities of the uterus, the possible tubar remnants, the vestigial lamina and the ovaries.

➢ Biological status - The karyotype of MRKH patients is always 46, XX with no visible chromosome modification.

➢ Spine radiography.

➢ Audiogram.

➢ Heart echography.

Treatment :

- Creating a neovagina only when they are ready to start sexual activity & emotionally mature.
- Treatment may be either surgical or nonsurgical.
- There are two main types of procedure.
- The first one consists of the creation of a new cavity and can be nonsurgical (Franck's dilator method) or surgical (Abbe-McIndoe/ Vecchietti/ Sigmoidal colpoplasty)
- The second is vaginal replacement with a pre-existing canal lined with a mucous membrane (a segment of bowel).
- Nonsurgical creation of a neovagina should be the first-line approach, if suitable.
- When a surgical approach is chosen, the surgeon must be experienced with the procedure.

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