INTRODUCTION

The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome or Mullerian duct agenesis is characterized by congenital aplasia of the uterus and the upper part (2/3) of the vagina in women showing normal development of secondary sexual characteristics and a normal 46, XX karyotype. Till date, there are only four case reports of MRKH syndrome with alopecia. The presence of alopecia with MRKH syndrome is a mere coincidence or an associated finding is a subject of research and may require further evidence.

CASE REPORT

A 17-year-old girl presented with hair loss from the scalp for 1 year. She also had a history of primary amenorrhea. The loss of hair begins at the occipital area of the scalp, and it progressed along the lateral margins of the scalp in a band like fashion clinically suggestive of ophiasis type of alopecia areata. The follicular openings were well preserved with normal skin texture [Figure 1a]. Detailed gynecological examination revealed normal secondary sexual characters with well-developed external genitalia and hypoplastic vagina [Figure 1b]. The histopathology of the scalp revealed lymphocytic infiltration around the lower third of the hair follicle, a finding suggestive of alopecia areata. To rule out the cause of amenorrhea, ultrasonography of abdomen and pelvis was done which revealed the absence of uterus and right kidney with normal ovaries [Figure 2a and b]. Hormone levels (follicle-stimulating hormone, luteinizing hormone, estradiol, testosterone, and thyroid function test) were within normal limits. The karyotyping showed 46 XX.

DISCUSSION

The detailed review of four case reports of MRKH syndrome with alopecia in world’s literature has been given in Table 1. First, three reports were from the same geographical region of Middle East (Jordan, Lebanon, and Turkey, respectively). Megerbane et al. and Tatar et al. hypothesized that the founder mutation in Middle East population might be responsible for the condition to
be restricted to that geographical region. However, the fourth case report from South Asia (Pakistan) questioned the hypothesis of founder mutation leading to MRKH syndrome restricted to that geographical region.\textsuperscript{[4]} We are reporting the fifth case report which is the second case report from South Asia and first among Indian population. Our case report is another evidence to suggest that MRKH syndrome with alopecia is not restricted to Middle East population.

In all previously reported four case reports, there was a history of parental consanguinity and siblings were also affected with MRKH syndrome and alopecia. In our case, there was no history of parental consanguinity and siblings were normal. There are many case reports and syndromes of hypogonadism with alopecia.\textsuperscript{[5-7]} In all the previous case reports of MRKH syndrome with alopecia, hypergonadotropic hypogonadism was noted. This case is the first report of MRKH syndrome with alopecia with normal gonadal function. MRKH syndrome may have associated abnormalities such as renal agenesis, skeletal abnormalities, hearing loss, or cardiac defects.\textsuperscript{[8,9]} In our case, the right renal agenesis was the associated finding of MRKH syndrome with alopecia which is not observed in all the previous four case reports under review.

In a female patient of alopecia areata, history of primary amenorrhea may be important to rule out the rare association with MRKH syndrome. This syndrome is caused by embryologic growth failure of the Mullerian duct with resultant agenesis or underdevelopment of the vagina, uterus or the both. All the reported cases of MRKH with alopecia including our case had alopecia areata which is an autoimmune

Table 1: Review of literature of MRKH syndrome with alopecia

| Authors                  | Siblings | Consanguinity | Karyotype | Uterus | Fallopian tubes | Ovaries | Alopecia | Other abnormalities          |
|--------------------------|----------|---------------|-----------|--------|----------------|---------|----------|-----------------------------|
| Al Awadi et al. (1985)\textsuperscript{[1]} | 2 sisters | Yes           | 46 XX     | Hypoplastic | One absent another hypoplastic | One D   | Partial alopecia consisting of cranial hair only in the center of scalp (Alopecia areata). Eyelashes and eyebrows normal. Sparse axillary and pubic hair | Microcephaly, flat occiput |
| Megarbane A et al. (2009)\textsuperscript{[2]} | 2 sisters | Yes           | 46 XX     | Hypoplastic | Hypoplastic     | A       | Partial alopecia. Sparse eyebrows. | Mild mental retardation, microcephaly, flat occiput |
| Tatar A et al. (2009)\textsuperscript{[3]} | 2 sisters | Yes           | 46 XX     | Hypoplastic | Hypoplastic     | A       | Partial alopecia. Sparse eyebrows. | Hypoplastic vagina |
| Zaman and Nisar (2009)\textsuperscript{[4]} | 2 sisters | Yes           | 46 XX     | One absent another rudimentary | Hypoplastic | A       | Alopecia totalis. Eyebrows absent. Eyelashes normal. Axillary and pubic hair sparse | Hypoplastic vagina |
| Our case (2016)          | Other sibling (male) is normal | No          | 46 XX     | Absent   | Absent         | N       | Alopecia areata with ophiasis pattern. Normal eyebrows, eyelashes, axillary and pubic hair | Right renal agenesis |

A: Agenesis, D: Dysgenetic, N: Normal

Figure 1: Clinical findings shows (a) ophiasis type of alopecia areata, (b) well-developed external genitalia and hypoplastic vagina

Figure 2: Ultrasonography of abdomen and pelvis shows (a) absence of uterus, (b) absent right kidney
disease. There seems to be no direct correlation between these entities as one is due to a genetic defect, and other is an autoimmune disease. Hence, the presence of alopecia in the case of MRKH syndrome is a mere coincidence, or an associated finding of the syndrome is a matter of further study.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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