The MURCS Association – Mullerian Aplasia, Renal Agenesis, Cervicothoracic Somite Dysplasia

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ABSTRACT

The MURCS (Mullerian agenesis, unilateral renal agenesis and cervicothoracic somite deformity) syndrome is a rare disease presenting with normal secondary sexual characters but uterus and upper vagina are absent due to mullerian agenesis. They usually present with primary amenorrhea. This case is unique because her primary diagnosis was chronic kidney disease and she was spotted in the dialysis unit. On thorough examination, her background diagnosis was arrived at. This case illustrates the fact that every patient must be examined in detail clinically.

Keywords: Fetal anomalies, renal failure, mullerian agenesis, dialysis, chromosomes

Embryogenesis and fetal development is a crucial period of life. Proper nutrition and care is a must to ensure a good outcome for the fetus. The care begins from the day of conception and any mistake afterwards will take its toll. Lack of care for even a single day can give rise to permanent damage in future. Here we will talk about one such condition in which teratogenicity occurs during end of 4th week of intrauterine life (Fig. 1). Let's see what happens next.

METHODS AND STATISTICS

A single case study is presented here as a rare case.

CASE STUDY

An 18-year-old female patient who presented with complaints of generalized weakness, easy fatigability, breathlessness on exertion, generalized anasarca and decrease in urine output was admitted. Patient had no complaints of decrease in appetite, decrease in food intake, difficulty in swallowing, yellow urine or sclera, abdominal distension, chest pain, perspiration, fever, sore throat, cough, cold, burning micturition or bleeding from any site. Patient was asked about menstrual history to rule out heavy bleeding recently or few months back but on the contrary, patient gave history of primary amenorrhea till date. Patient had no similar episodes in past.

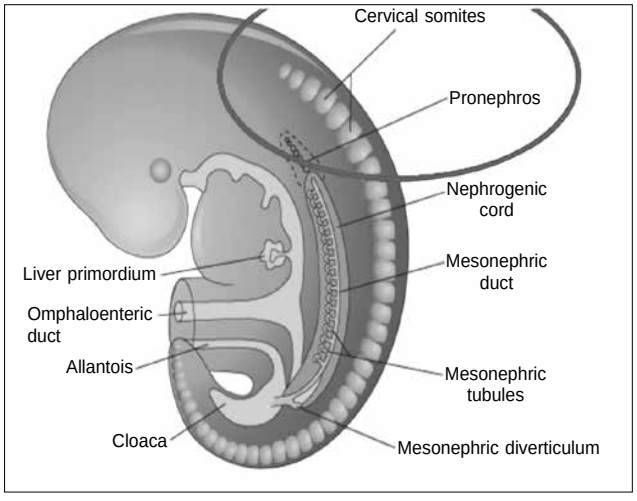

Figure 1. 4th week of intrauterine life.

On examination, patient was conscious and oriented to time-place-person. She had short stature, mild tachycardia with hyperdynamic pulse, hypertension, severe pallor without apparent jaundice and generalized edema with periorbital swelling and pitting pedal edema. Lung fields were clear with no heart murmurs. No organomegaly or tenderness observed over abdomen. Per vaginally, patient had small vaginal opening which on further examination was found to be abruptly

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ending in short course. Patient had normal secondary sexual characteristics. Blood picture showed decrease in hemoglobin to 5.3 g/dL with severe microcytic hypochromic anemia with normal bilirubin levels. Urine examination showed albuminuria, hematuria and pyuria. Renal function tests showed creatinine value of 24.5 mg/dL and urea of 287 mg/dL. Ultrasonography of abdomen and pelvic region revealed unilateral single kidney with altered echotexture, absent uterus and absent upper two-third part of vagina, suggestive of mullerian agenesis. Ovaries were normal. To rule out other complications, X-ray of cervicothoracic spine was done which revealed scoliosis. A diagnosis of MURCS association (Mullerian agenesis, unilateral renal agenesis and cervicothoracic somite deformity) was made. Barr body test came negative and further karyotyping done to assess any chromosomal abnormality showed normal chromosome structure of 46, XX. Patient was managed by hemodialysis with blood transfusion and antibiotics for urinary infection, antihypertensive, nutritional supplementation especially iron sucrose and erythropoietin, dietary interventions, etc. Later on, an arteriovenous fistula was inserted for maintenance dialysis. Patient was taught about gradual dilatation technique of vagina and explained about the disease and its prognosis. Echocardiography was done to rule out cardiac anomalies, which was normal.

DISCUSSION

MURCS association is a syndrome characterized by mullerian aplasia or hypoplasia, unilateral renal agenesis or ectopy and cervicothoracic somite dysplasia which leads to defects in vertebra (e.g., scoliosis in this case), ribs, upper limbs and scapula. Other anomalies can also be seen, such as anorectal, cardiac, pulmonary and ovarian anomalies. The pathophysiology is not clear but it is believed to be an event occurring very early in development around end of 4th week of intrauterine life when the blastemas of pronephric buds and cervicothoracic buds are closely located. Regarding pathogenesis, it is hypothesized that there can be possibility of an unidentified teratogen as there is no chromosomal abnormality or familial transmission. The disease has similarities with 22q11 deletion syndrome and Mayer-Rokitansky-Küster-Hauser syndrome, suggesting a similar pathophysiology. Usually, the patient undergoes normal thelarche but menarche is the stage where the patient seeks help. Secondary sexual characteristics and bodily changes associated with menstruation are normal because ovaries are normal. Infertility, difficult voiding, skeletal abnormality, difficult intercourse, etc. could be the presentation. Per vaginal examination can be difficult. Diagnosis along with screening for all possible anomalies require ultrasonography, X-ray chest and cervicothoracic spine, magnetic resonance imaging (MRI), laparoscopy, pyelography, echocardiography, karyotyping, serum follicle stimulating and luteinizing hormone estimation, testosterone levels, etc. The syndrome is managed by frank perineal dilatation technique and various surgeries for reconstruction of vagina. Conception can happen with assisted reproductive techniques. Having a genetic offspring is possible through a gestational carrier. Psychological counseling is needed for gender identity issues if present. Other anomalies should be corrected e.g., renal replacement therapy for renal anomalies, orthopedic repair of vertebral anomalies, cardio surgery, etc.

CONCLUSION

As we can see, this association between anomalies in this syndrome requires a combined approach from almost all specialties namely, gynecologist, physician, general surgeon, obstetric specialist, nephrologist, orthopedic surgeon, pathologist, microbiologist, biochemist, endocrinologist, cardiologist, cardio surgeon, pediatrician, psychiatrist, ENT surgeon, etc. Moreover, patient's willingness and effort is also required.

"I can do things, you cannot;
You can do things, I cannot;
Together we can do great things."

Although this may seem like too much of work, at the same time we should not forget our oath because:

"Helping one person might not change the whole world,
But it could change the world for one person."

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