A Turner syndrome case associated with anal atresia, interrupted aortic arch and multicystic dysplastic kidney

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Although renal and cardiac malformations are commonly seen in Turner syndrome (TS), anorectal malformations, multicystic dysplastic kidney and interrupted aortic arch are quite rare in TS. A newborn with TS with three quite rare congenital malformations (imperforate anus/anal atresia associated with rectovestibular fistula, interrupted aortic arch, and multicystic dysplastic kidney) is presented.

Key words: Turner syndrome, imperforate anus, anal atresia, multicystic dysplastic kidney, interrupted aortic arch.

Case Report

A 2800 g (25th to 50th percentile) female newborn was born at term by cesarean section. Her length was 46 cm (3rd to 10th percentile), and her Apgar scores at the first and fifth minute were eight and nine, respectively. Fetal ultrasonography showed an increase in nuchal translucency and left multicystic dysplastic kidney at the fourth month. Amniocentesis was performed in the 18th week of pregnancy and 45,X was observed in all fields. Therapeutic abortus was suggested to the family, but was not accepted. On physical examination, she had low posterior hairline, short neck, low-set ears, capillary hemangioma on nose and left eyelid, widely spaced nipples, excess loose skin on the back of the neck, and imperforate anus (Fig. 1). A continuous murmur was heard. Echocardiography showed IAA and patent foramen ovale. Descending aorta was watered via patent ductus arteriosus and she had severe pulmonary hypertension. Left multicystic dysplastic kidney was confirmed by postnatal ultrasonography (Fig. 2). Vesicoureteric reflux (VUR) was not detected. No sacral agenesis or hemivertebra was observed on radiographic investigations.
Discussion

Turner syndrome (TS) is one of the most common sex chromosomal aberrations, and may be associated with various congenital malformations. Major congenital malformations such as of the cardiovascular and genitourinary systems are more frequently observed in TS with non-mosaic 45,X karyotype\(^3\). When TS is diagnosed, associated malformations such as cardiac and renal malformations must be investigated. The incidence of cardiovascular malformations in TS is 14%-42\%\(^4\). Severe congenital cardiovascular malformations are more frequent in the 45,X karyotype\(^4\), as in our patient. The congenital cardiovascular malformations in TS usually consist of the vessels of the left side of the heart\(^4\). Bicuspid aortic valve and coarctation of the aorta are the most common forms of cardiovascular malformations. Dilated ascending aorta, hypoplastic aortic arch, mitral valve prolapse, interrupted inferior vena cava with azygos continuation, aortic valve disease, partial anomalous pulmonary venous return, ventricular septal defect, pulmonary valve abnormality, patent ductus arteriosus, and dextrocardia may also be seen in patients with TS\(^2,4,8\). IAA is relatively rare and usually associated with a ventricular septal defect and patent ductus arteriosus. In addition to IAA, patent ductus arteriosus and patent foramen ovale were observed in our case. Although IAA is commonly seen in DiGeorge syndrome, it is quite rare in TS. To the best of our knowledge, IAA associated with TS has been reported in only one case\(^9\). Our case is the second case of IAA and TS in the medical literature.

The frequency of renal malformation in children with TS is 33%-70\%\(^10,11\). Renal malformations are more common in patients with non-mosaic 45,X. Collecting system malformations are more frequently observed in patients with mosaic 45,X/46,XX\(^3\). Horseshoe kidney and abnormal positioning or duplication of renal pelvis, ureters or vessels are the most common kidney malformations in TS\(^3,4\), whereas multicystic dysplastic kidney is rare. Incidence of cystic kidney disease in TS is 1.8\%\(^12\). Multicystic dysplastic kidney is seen in 0.9\% of the cases with TS\(^12\). VUR in the contralateral kidney may be observed in TS with multicystic dysplastic kidney\(^12\). In our case, VUR was not determined.

Gastrointestinal pathologies such as intestinal telangiectasia\(^6\), colon carcinomas\(^13,14\), inflammatory bowel diseases\(^15\), situs inversus totalis\(^8\), elevated liver parameters\(^2,4\), achalasia\(^5\), cirrhosis\(^16\), duodenal angiodyplasia\(^17\), gastrointestinal bleeding\(^18\), and imperforate anus\(^7\) have been reported in association with TS. Although anorectal malformations are one of the most common congenital defects, they are quite rare in TS. As far as we know, only one case of Wilms tumor and imperforate anus was reported in a patient with TS\(^7\). Incidence of anorectal malformations is 1 per 4,000 live births\(^19\). It may be a component of VATER
or VACTERL association. Our case had renal, cardiovascular and anorectal malformations, but no vertebral, tracheoesophageal or limb malformations. The most common anorectal anomaly in females is anal atresia associated with a rectovestibular fistula, and the level of the anal atresia is usually intermediate. Our patient also had anal atresia associated with a rectovestibular fistula but its level was low. Tumors may develop in patients with TS such as Wilms tumor and colon cancer. For that reason, patients with TS should be closely followed for the risk of tumor development. Differential diagnosis should be made considering the other syndromes presenting malformations of the cardiovascular, genitourinary and gastrointestinal systems.

To our knowledge, the association of IAA, imperforate anus/anal atresia associated with rectovestibular fistula and multicystic dysplastic kidney in TS has not been reported previously. Congenital cardiac, renal and gastrointestinal system malformations should be investigated in newborns with TS. Early diagnosis and treatment of severe congenital malformations will prevent development of serious complications.

REFERENCES