The Turkish Journal of Pediatrics 28:85 - 89, 1986

BLADDER DISCONFIGURATION IN CONJUNCTION WITH THE VATER ASSOCIATED ANOMALIES*
(Say Syndrome)

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Key words: multiple congenital anomalies, Say syndrome, VATER association

Say and Gerald[1] first recognized that the triad of polydactyly, imperforate anus and vertebral anomalies might be regarded as a new syndrome and then Quan and Smith[2] described the VATER association. The letters represent the anomalies involved, V: vertebral, A: anorectal, TE: tracheoesophageal, R: radial and renal anomalies[1-4], which are found together in a non-random manner. The reports that followed extended the scope of the anomalies. Congenital heart disease, anomalies of external genitalia, intestinal anomalies, single umbilical artery, tracheal atresia and esophageal muscular ring have been described as additional anomalies in the VATER association[5-7].

Case Report

The one-day-old patient was the first child of a 14-year-old mother and a 17-year-old father; the parents were not related. There were no recognizable teratogenic factors in the pregnancy and no family history of congenital anomalies. The baby was born at home by spontaneous vertex delivery at 40 weeks of gestation and weighed 2600 grams. The complaints were the absence of anus and failure to swallow. On examination esophageal atresia, bilateral radial aplasia, anorectal anomaly and glandular hypospadias were found. Radiological examinations showed proximal atresia of the esophagus with distal tracheoesophageal fistulas, cervical hemivertebrae, the infralevator type anorectal anomaly without fistula and bilateral absence of ossa radii while a retrograd cystogram revealed bladder disconfiguration (Figures 1, 2). Chromosome analysis showed a normal male karyotype.

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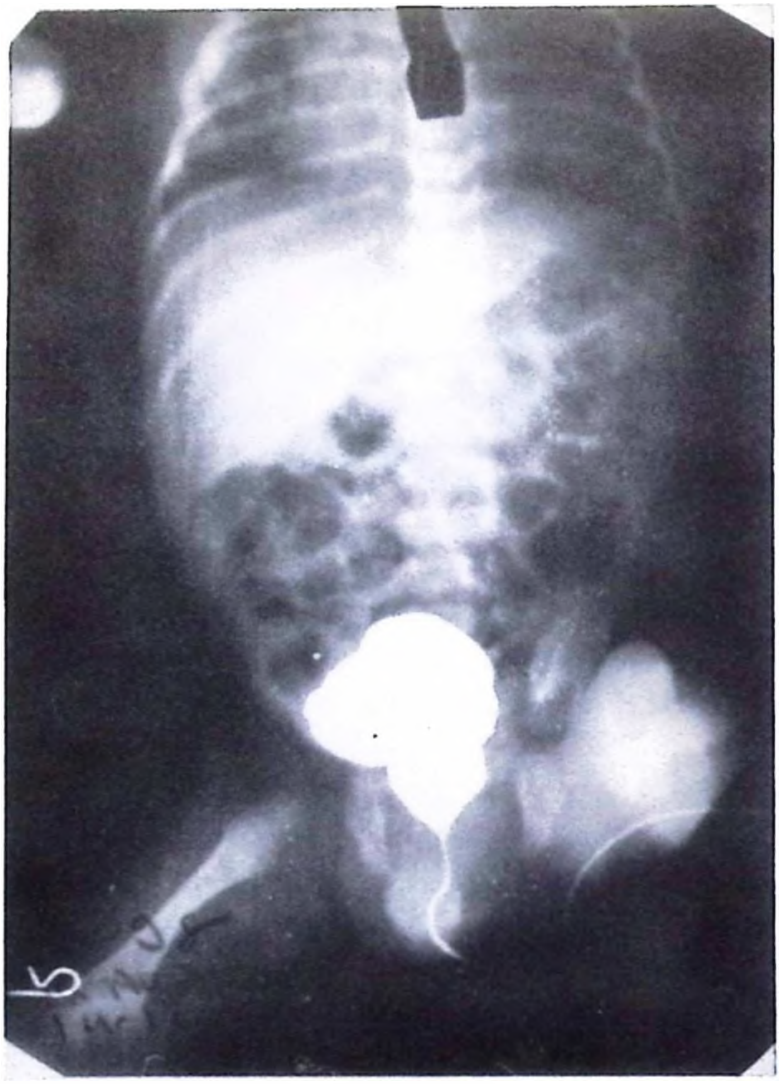

Figure 1

Anteroposterior view of the bladder.

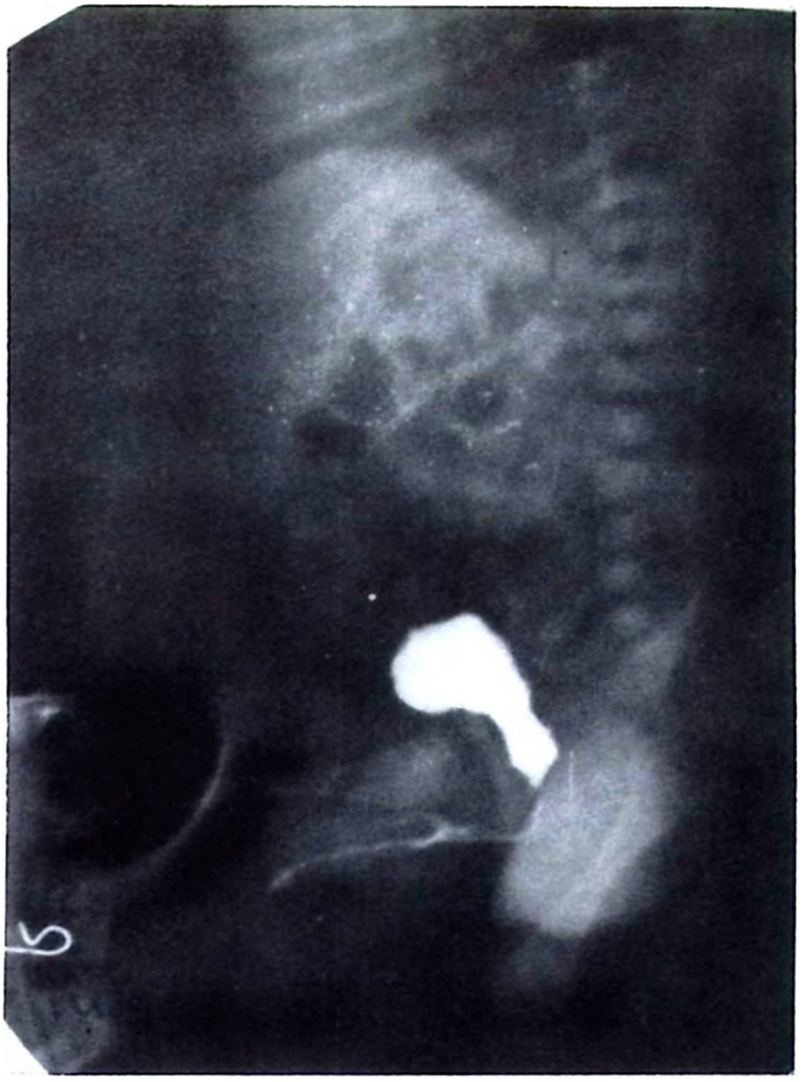

Figure 2
Right side of the bladder.

On admittance, a perineal anoplasty was carried out and after the patient had been examined for further associated anomalies, the tracheosophageal fistulas were separated and a primary anastomosis to the esophagus was performed. After an uneventful recovery the patient was discharged but anal dilatations and urologic controls were advised. After discharge, he had an attack of bronchopneumonia. He was brought back for a check-up at the 3rd postoperative month with the complaint of failure to thrive.

Discussion

It is agreed that the VATER association does not describe a single etiologic syndrome, but a tendency for certain anomalies to present together. A common type of defect in the mesoderm occurring between the 4th and 7th weeks of intrauterine life is thought to be the basis for such an association[4,5,8,9]. Only one family history of occurrence within a sibship has been reported to date[10]. No responsible teratogens and no chromosomal abnormalities have been detected as causative agents.

The term VATER association is important as a memory device. The presence of one of the associated anomalies should alert the physician to investigate for the other anomalies, because prompt diagnosis is important if we are to achieve long-term meaningful survival for these patients as is often possible[11,12]. All systems should be thoroughly investigated, including cystograms. These investigations may reveal additional anomalies, even ones previously unreported as in our case where there was bladder disconfiguration.

Summary

The case of a newborn with the VATER associated defects, and a previously unreported additional anomaly, bladder disconfiguration, is reported. The importance of thorough investigations in the presence of one of the associated anomalies is stressed.

Acknowledgement

We would like to thank Prof. Dr. Sevim Balcı of the Hacettepe University Institute of Child Health for kindly undertaking the chromosome analysis.

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