An intriguing co-occurrence of MURCS and VACTER associations

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Background

MURCS and VACTER associations have several defects in common, and yet they are considered distinct clinical entities. In both, the underlying cause is still unknown.

MU
- Mullerian agenesis

RC
- Renal anomalies

C
- Cervicothoracic-

S
- Somite abnormalities

V
- Vertebral defects

A
- Anal atresia

C
- Cardiac defects

TE
- Tracheo-Esophageal fistula

R
- Renal anomalies

Methods

- Pelvic ultra-sound and MRI
- Cervical X-ray
- Genetic testing
  - Karyotype
  - Chromosomal Microarray Analysis
- Review of the literature

Patient

A 12 year old girl who was referred for genetic counseling

- Tracheo-esophageal fistula
- Fusion of the labia minora
- Short stature

Results

- Vaginal atresia
- Normal karyotype and normal CMA
- Horseshoe kidney
- C6-7 blocked vertebrae

Conclusions

- This patient meets the diagnostic criteria of both MURCS and VACTER associations.
- A co-occurrence of the two has been reported in only 3 case reports in the past.
- Future studies will hopefully reveal the embryonal and genetic mechanism leading to these congenital defects.
- Discovering the underlying cause will enable accurate follow-up and genetic counseling regarding recurrence risk.