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
- **R** •Renal anomalies
- **C** •Cervicothoracic-
- **S** •Somite abnormalities

- **V** •Vertebral defects
- **A** •Anal atresia
- **C** •Cardiac defects
- **TE** •Tracheo-Esophageal fistula
- **R** •Renal anomalies

Patient

A 12 year old girl who was referred for genetic counseling

Tracheo-esophageal fistula

Fusion of the labia minora

Short stature

Methods

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

Results

C6-7 blocked vertebrae

Vaginal atresia

Normal karyotype and normal CMA

Horseshoe kidney

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.

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