

## Megalopenis in an infant with VACTERL association

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Complete spectrum of VACTERL association comprises vertebral anomalies, anal atresia, congenital cardiac defects, oesophageal atresia, renal dysplasia and limb anomalies<sup>1</sup>. Prune-belly syndrome is characterized by the triad of deficiency of the anterior abdominal wall muscles, dilatation of urinary tract and cryptorchidism<sup>2</sup>. Three cases of concordance of VACTERL association and prune-belly syndrome have been reported<sup>3,4,5</sup>. We report a case of VACTERL association who also has megalopenis and most of the features of prune-belly syndrome.

### Case Report

A 4-month-old infant was referred from a surgical unit for the evaluation of a febrile illness. He was the first child of non-consanguineous parents. There was oligohydroamnios antenatally and the child was born at 38 weeks of gestation by elective caesarean section due to transverse lie. The fetal movements had been normal.

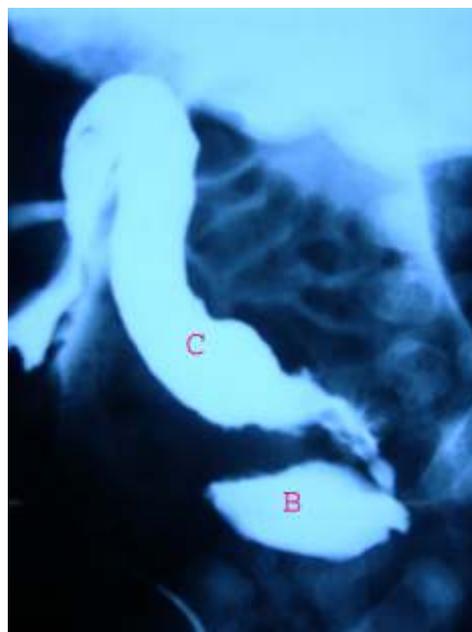


**Figure 1** Sacral agenesis (>), absent gas in distal large bowel (<<)

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Soon after birth, a high imperforate anus was detected (figure 1) and sigmoid colostomy was done on the second day of life. Furthermore, distal loopogram showed recto-vesical fistula (Figure 2).



**Figure 2** Distal loopogram showing recto-vesical fistula. Colon(C), Bladder (B)

Further investigations revealed that the patient had sacral agenesis, ostium secundum atrial septal defect, patent ductus arteriosus and a small hydronephrotic left kidney with poor cortico-medullary demarcation. The bladder wall was thickened and ureters were not dilated. This patient did not have tracheo-oesophageal fistula. On the basis of above findings a diagnosis of VACTERL association was made.

The most striking feature of this child was his large phallus (megalopenis). It was 7.5cm in length and the overlying skin of the penis had rugosities (figure 3, 4). There was bilateral cryptorchidism. Investigations revealed that both testicles were in the inguinal canals, penis was hypertrophied and urethra was dilated (megalourethra). The anterior abdominal wall was lax though deficiency of the abdominal musculature could not be reliably excluded. These findings suggested an incomplete prune-belly syndrome.



**Figure 3 & 4** Megalopenis with overlying skin rugosities

### Discussion

Associations such as VACTERL are caused by nonspecific developmental disruption acting on developmental field (embryo) in early first trimester<sup>6</sup>. A disruption in differentiating mesoderm in first 4-5 weeks has been suggested to be the basis for VACTERL association. Hence, other mesodermal structural defects are occasionally found with VACTERL association<sup>7</sup>. Urogenital anomalies like unilateral or bilateral cryptorchidism, hypospadias and micropenis in males and ambiguous genitalia and bladder exostrophy in females have been frequently described in VACTERL association<sup>8,9</sup>. In the largest such series of 286 patients, 81 (28%) had severe genital defects<sup>7</sup>. Abdominal wall abnormalities like omphalocele and gastroschisis are also occasionally reported.

As far as we know our case is the second reported case of megalopenis associated with VACTERL association. First case was reported in India by Shah D *et al*<sup>4</sup>. In this case, complete form of prune-belly syndrome concordance with VACTERL association was seen. The patient had megalopenis with rugosities of the overlying skin as in our case.

Lukusa, *et al*<sup>5</sup>. described incomplete prune belly anomaly in a female child with additional features of the VACTERL association. Ozturk, *et al*<sup>3</sup> for the first time reported concordance of complete prune belly syndrome and VACTERL association in a premature male child. Our case had bilateral cryptorchidism, lax abdominal wall and megalourethra. These features would suggest the additional diagnosis of prune-belly syndrome. Early disturbance of mesodermal development in

both the abdominal wall and the urinary tract have also been suggested to be responsible for prune belly syndrome<sup>10</sup>. This should explain why VACTERL association and prune-belly syndrome sometimes occur in concordance.

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