

Case-Based Diagnosis Training

Patient:

Gender: *Female*

Age: *1 day old*

Clinical history and working diagnosis on the referral:

Newborn 38 5/7 AOG via repeat CS, small for gestational age

Working diagnosis: VACTERL association

Normal variant:

Apex beat at the right, shallow left nasolabial fold, imperforate anus, overly flexed upper extremities, absent movement of the left thumb, decreased movement of the 2nd and 3rd digits of the left hand, wrist drop at the left hand, sacral dimpling

Submitted by:

M.M. Cariño

Please add pictures (radiograph, ultrasound, CT or MR images) and schematic drawing of the developmental process *if applicable* by clicking on the symbols within the boxes below:



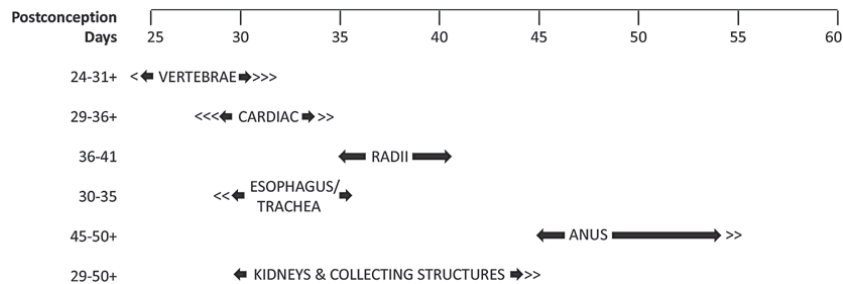
Picture 1-2: Babygram (PA and lateral views)

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Additional information

In case you want to submit further pictures, please add these (radiograph, ultrasound, CT or MR images) and schematic drawing of the developmental process *if applicable* by clicking on the symbols within the boxes below:

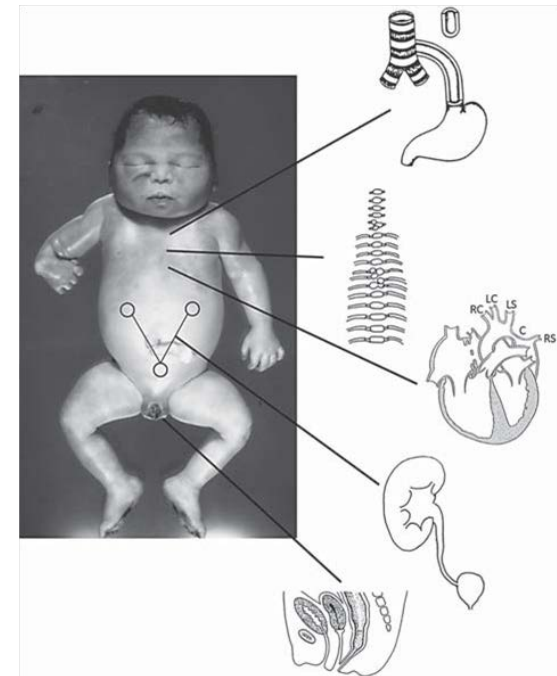
Underlying step in embryological development:



Reference: Stevenson RE, Hunter AGW. Considering the Embryopathogenesis of VACTERL Association. *Molecular Syndromology*. 2013;4(1-2):7-15. doi:10.1159/000346192

Potential differential diagnostic entities:

- CHARGE syndrome
- Holt Oram syndrome
- DiGeorge syndrome
- Townes-Brocks syndrome
- Trisomy 18



Schematic drawing of the developmental abnormalities in VACTERL syndrome. Reference: Stevenson RE, Hunter AGW. Considering the Embryopathogenesis of VACTERL Association. *Molecular Syndromology*. 2013;4(1-2):7-15. doi:10.1159/000346192.

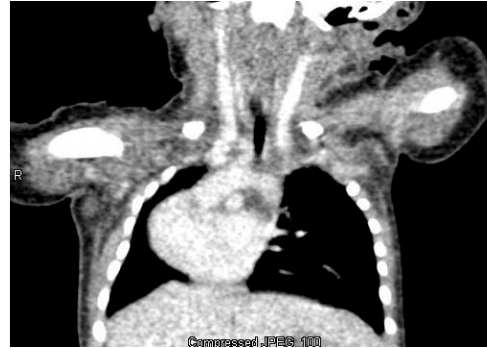
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Additional pictures

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Picture 5: Dextrocardia



Picture 6: Right-sided aortic arch



Picture 7: Persistent left superior vena cava draining to the right atrium



Picture 8: Pulmonary artery sling



Picture 9: Mild to moderate narrowing of the mid to lower trachea to approx 49%



Picture 10: Aberrant left pulmonary artery arising from the right pulmonary artery

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Additional pictures

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Fusion of L1 and RIGHT L2 hemivertebra, Fusion of LEFT L2 hemivertebra and L3, Fusion of L4 & L5

Crossed fused renal ectopia