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| **Part 1 of 3 - Case 1** |
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| **Question 1** | 1.0 Points |

A 9-day-old term 3.2 kg female neonate delivered via vaginal birth at 39 weeks presented with cyanosis. A physical exam found no external abnormalities at the umbilicus, genitalia, anus, spine, or lower limbs. However, the child’s hands exhibited transverse palmar creases as shown in figure 1a, and the child’s facial features included epicanthal folds, slanted palpebral fissures, and flat nasal bridge as shown in figure 1b -note that that the jaw, upper lip, and philtrum are normal. A review of mother's records showed no indication of either oligo- or polyhydramnios during the pregnangy. Transesophageal echocardiography demonstrated abnormal flow between the atria and ventricles as well as the presence of atrial and ventricular septal defects.The external features of the child are **MOST CONSISTENT** with which of the following?

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| CorrectA. Down syndrome |  |
| IncorrectB. trisomy 18 |  |
| IncorrectC. VACTERL association |  |
| IncorrectD. Fetal Alcohol Syndrome |  |
| IncorrectE. Potter sequence |  |

Feedback: The facial and palmar features are **MOST** consistent with Down syndrome (trisomy 21). Trisomy 18 often presents with low-set ears and reduced mandible. Fetal Alcohol Syndrome is correlated with the following discriminating features, all of which were NOT observed in this case:* short palpebral fissures (small eye openings)
* reduced upper lip
* smooth philtrum

VACTERL is an entire spectrum of findings including:* **V**ertebral defects
* **A**nal atresia
* **C**ardiovascular defects
* **T**racheo-**E**sophageal defects
* **R**enal defects
* **L**imb defects

While there are certainly cardiovascular and 'limb' defects (insofar as a palmar crease may be considered a defect), there is NOT any evidence to suggest the presence of other elements within the VACTERL association.**Question 2** |

Which if the following **BEST** explains the entire spectrum of cardiovascular defects observed in Case 1?

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| IncorrectA. failure of the atrial septum primum to form |  |
| IncorrectB. failure of the muscular ventricular septum to form |  |
| CorrectC. failure of the atrioventricular cushions to fully develop |  |
| IncorrectD. failure of cardiac neural crest to migrate or fully develop  |  |

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Feedback: The defect described is most consistent with a failure of the atrioventicular cushions to fully develop and the atrioventricular canal to NOT be properly divided into left and right A-V canals. Under these conditions, you would expect to see defects in the inferior atrial septum AND superior ventricular septum (membranous septum) as well as defects in the mitral and tricuspid valves. Recall that the A-V cushions are NOT neural crest derived.

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| **Part 2 of 3 - Case 2** |

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| **Question 3** | 1.0 Points |

A 19-year-old male is referred to a neurologist for evaluation of what appears to be a peripheral neuropathy. The patient's chief complaint is that his right arm is often cold with occasional numbness, weakness, and parasthesia on the extensor and flexor surfaces of the arm, forearm, and hand. He also reports claudication with heavy use, which is often accompanied by dizziness. A complete neurological exam reveals no other peripheral motor or sensory findings and intact cranial nerves, although the patient does report occasional difficulty swallowing. Physical findings reveal normal heart and lung sounds. Blood pressure readings taken on the right arm are consistently lower than those taken on the left, and the right radial pulse is noticeably weaker.The **MOST LIKELY** reason for this constellation of findings is:

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| A. aberrant origin of the right subclavian artery |  |
| B. ulnar nerve compression |  |
| C. pulmonary stenosis / Tetralogy of Fallot |  |
| D. coarctation of the aorta |  |
| E. vitamin B12 deficiency |  |

Feedback: The signs and symptoms are most consistent with disruption of blood flow to the right arm as would be caused by abnormal origin of the right subclavian artery from the aortic arch distal to the origin of the left subclavian artery. In this instance, the right subclavian artery courses behind the esophagus thus compressing the esophagus against the aorta and causing dysphagia as well as disrupting flow in the aberrant subclavian artery thus cuasing the symptoms seen in the right arm. Incorrect Feedback: The signs and symptoms are most consistent with disruption of blood flow to the right arm as would be caused by abnormal origin of the right subclavian artery from the aortic arch distal to the origin of the left subclavian artery. In this instance, the right subclavian artery courses behind the esophagus thus compressing the esophagus against the aorta and causing dysphagia as well as disrupting flow in the aberrant subclavian artery thus cuasing the symptoms seen in the right arm. |
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| **Question 4** | 0.0 Points |

**BONUS:** Note that during exertion of the right arm, vessels supplying the muscle will open up in an attempt to provide adequate blood to the muscle. However, the patient reports significant pain suggesting that despite this vasodilation, there is insufficient blood flow to sustain exertion. Moreover, the patient also reports that he becomes dizzy when this happens suggesting that the flow of blood to the brain is being reduced. Briefly explain what might be going on to cause this. (Free text response)

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| **Part 3 of 3 - Case 3** |

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| **Question 5** | 1.0 Points |

A young couple who have adopted a 11-month-old boy with a known cardiovascular condition are referred to a pediatric cardiologist for the child's care. The child is generally cyanotic on any exertion, tachypnic at rest and frequently adopts a squatting posture with his legs pressed against his chest. Transesophageal echocardiography shows the presence of pulmonary stenosis, right ventricular hypertrophy, and a membranous ventricular septal defect. On physical exam, the child is alert but lethargic and exhibits facial dysmorphology (see figure) including reduced palpebral fissures, reduced upper lip, smooth philtrum, low-set ears, and a head-circumference in the 50th percentile. The cardiovascular findings are **MOST CONSISTENT** with which of the following?

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| IncorrectA. coarctation of the aorta |  |
| CorrectB. tetralogy of Fallot |  |
| IncorrectC. persistent atrioventricular canal |  |
| IncorrectD. transposition of the great vessels |  |
| IncorrectE. hypoplastic left ventricle |  |

Feedback: The findings listed include three which are associated with tetralogy of Fallot:* pulmonary stenosis
* ventricular septal defect (VSD)
* right ventricular hypertrophy

More likely than not, more careful imaging would show that the opening of the aorta is centered over (over-riding) the VSD.  In addition, the ductus arteriosus is likely still patent to allow shunting of blood to the lungs. |
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| **Question 6** | 1.0 Points |

Which if the following **BEST** explains the entire spectrum of cardiovasculardefects observed in Case 3?

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| A. failure of the atrial septum primum to form |  |
| B. failure of the muscular ventricular septum to form |  |
| C. failure of the atrioventricular cushions to fully develop |  |
| D. failure of the cardiac neural crest to migrate and/or fully develop |  |

Feedback: Recall that the outflow tract is divided into pulmonary and aortic trunks in a spiraling fashion such that the pulmonic trunk is associated with the right ventricle and the aorta is associated with the left ventricle and that this development is dependent on endocardial cushions in the outflow tract that arise from cardiac neural crest. In addition to dividing the outflow tract, these cushions also contribute to the formation of the membranous ventricular septum thus explaining the VSD observed.Incorrect Feedback: Recall that the outflow tract is divided into pulmonary and aortic trunks in a spiraling fashion such that the pulmonic trunk is associated with the right ventricle and the aorta is associated with the left ventricle and that this development is dependent on endocardial cushions in the outflow tract that arise from cardiac neural crest. In addition to dividing the outflow tract, these cushions also contribute to the formation of the membranous ventricular septum thus explaining the VSD observed. |
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| **Question 7 of 7** | 1.0 Points |

The external features of the child are **MOST CONSISTENT** with which of the following? |
| IncorrectA. Down syndrome |  |
| IncorrectB. trisomy 18 |  |
| IncorrectC. VACTERL association |  |
| CorrectD. Fetal Alcohol Syndrome |  |
| IncorrectE. Potter sequence |  |

Feedback: Fetal Alcohol Syndrome is correlated with the following discriminating features, all of which are observed in this case:

* short palpebral fissures (small eye openings)
* reduced upper lip
* smooth philtrum