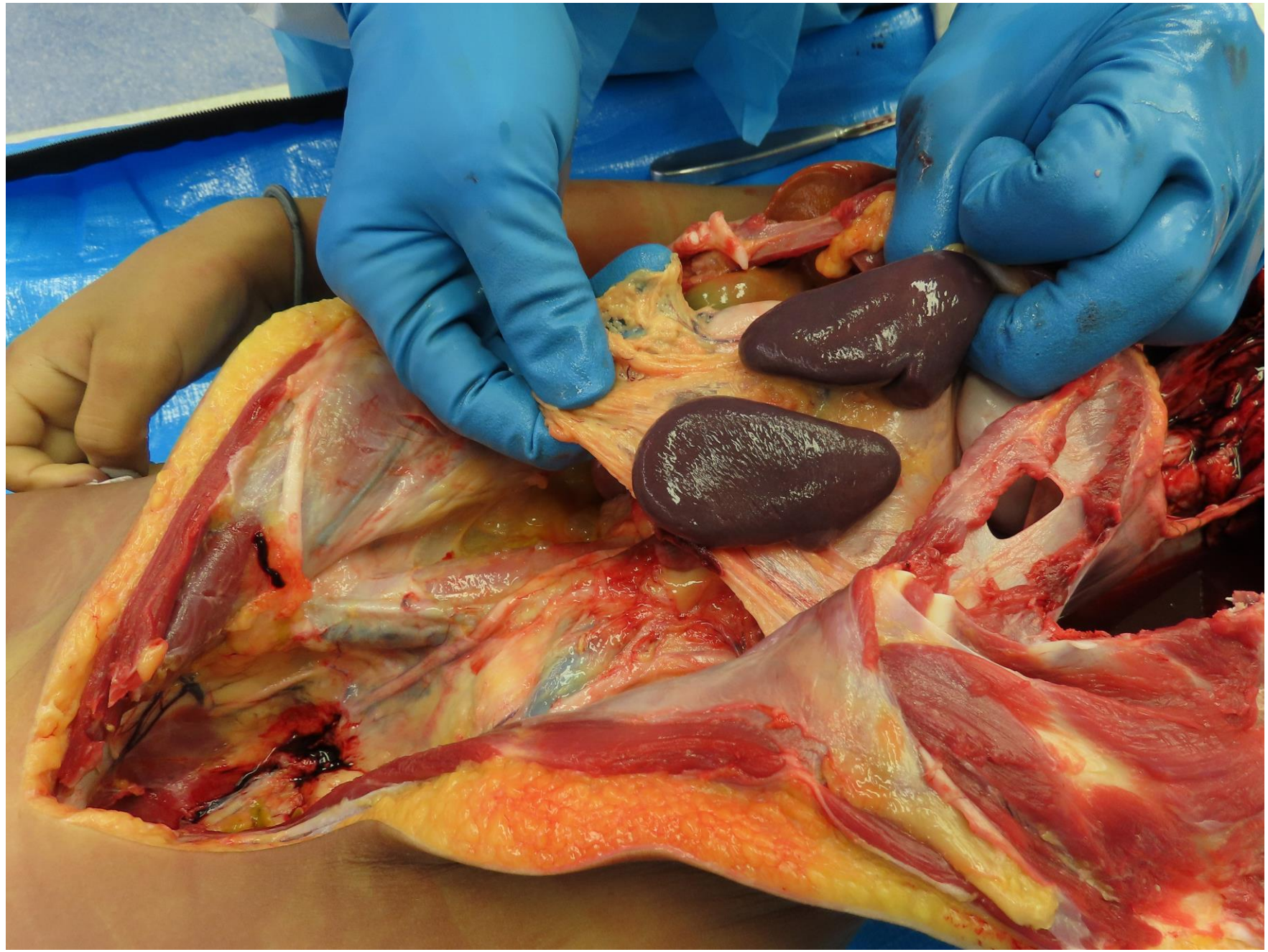
Case #97

NAME Educational Activities Committee

Case provided by:

Dr. Fabiola Righi (Forensic Pathology Fellow, Dallas County).



1. The decedent is a 7-year-old male with no known medical history, and recent complains of fever and cough. He is found unresponsive in bed by family. During autopsy, the pictured finding was identified.

What is commonly associated with this finding?

- ◯ Congenital heart defects
- ◯ DiGeorge syndrome
- ◯ Neural tube defects
- ◯ Trisomy 21

Answer...

A. Congenital heart defects – (CORRECT ANSWER, 52.55 % of responses)

The findings of multiple spleens is often seen in the setting of with Polysplenia Syndrome, a rare congenital subtype of heterotaxia syndrome, usually characterized by left isomerism (bilateral left sidedness), associated with various cardiac anomalies, abnormal venous drainages and abdominal heterotaxia.

Various cardiovascular anomalies may be encountered with this syndrome including atrial isomerism, atrial and ventricular septal defects, bilateral superior vena cava, right-sided aortic arch, partial anomalous pulmonary venous return, transposition of the great arteries, pulmonary valvular stenosis, and subaortic stenosis.

Another thing to keep in mind is that sinus node dysfunction is a common cause of arrhythmia in patients with Polysplenia Syndrome. The development of the sinus node tissues is strongly related to the development of the right appendage. Therefore, in right isomerism two sinus nodes are usually found; conversely, a sinus node tends to be hypoplastic or absent in left isomerism, which is often seen with Polysplenia Syndrome.

Other responses…

B. DiGeorge Syndrome (33.78 % of responses)

DiGeorge syndrome, also known as 22q11.2 deletion syndrome, is related to an array of congenital malformations including distinct facial features, cleft palate, and congenital heart defects (such as ventricular septal defects and truncus arteriosus). Patients with DiGeorge syndrome are also have lowered immunity, hypoparathyroidism and are more prone to autoimmune disorders. DiGeorge syndrome is not commonly associated with polysplenia.

C. Neural tube defects (8.71 % of responses)

Neural tube defects (NTDs) occur when the neural tube does not close properly. The two most common NTDs are spina bifida (a spinal cord defect) and anencephaly (a brain defect). Common causes of NTDs include genetic factors, folic acid deficiency and other abnormal intrauterine environments in pregnancy. NTDs are often combined with other systemic malformations, most commonly urogenital. Association with polysplenia has not been commonly documented.

D. Trisomy 21 (4.95% of responses)

Trisomy 21, or Down Syndrome, is a genetic disorder that typically presents with mild to moderate intellectual disability, growth retardation, and characteristic facial features. It is also associated with numerous congenital anomalies, most commonly congenital heart defects (seen in approximately 50% of patients), followed by gastrointestinal atresias (usually duodenal or esophageal) and vertebral anomalies. Polysplenia is not typically associated with Trisomy 21.

REFERENCES

1. Akhtar F, Bokhari SRA. Down Syndrome. [Updated 2023 Aug 8]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2024 Jan-. Available from: https://www.ncbi.nlm.nih.gov/books/NBK526016/
2. El Mortaji H, Elatiqi K, El Hammaoui H, Alj S. Polysplenia syndrome with situs ambiguous, common mesentery, and IVC interruption discovered incidentally in an adult. Radiol Case Rep. 2019 Jun 29;14(9):1072-1075. doi: 10.1016/j.radcr.2019.05.032. PMID: 31320964; PMCID: PMC6612708.
3. Fukuda K, Onda T, Kimura Y, Miura S, Matsumori R, Masaki Y, Nishino A, Inoue K, Fujiwara Y, Sumiyoshi M. An adult case of polysplenia syndrome associated with sinus node dysfunction, dextrocardia, and systemic venous anomalies. Intern Med. 2015;54(9):1071-4. doi: 10.2169/internalmedicine.54.3354. Epub 2015 May 1. PMID: 25948350.
4. Hong, ZH., Jin, DH., Yuan, XJ. *et al.* Association of neural tube defects with congenital abnormalities of the urogenital system in a Chinese cohort. *BMC Pediatr* **21**, 66 (2021). https://doi.org/10.1186/s12887-021-02492-8
5. https://www.mayoclinic.org/diseases-conditions/digeorge-syndrome/symptoms-causes/syc-20353543#:~:text=people%20with%2022q11.-,2%20deletion%20syndrome.,behavioral%20and%20mental%20health%20problems.
6. https://www.hopkinsmedicine.org/health/conditions-and-diseases/neural-tube-defects
7. https://www.cdc.gov/ncbddd/birthdefects/surveillancemanual/quick-reference-handbook/trisomy-21-down-syndrome.html#:~:text=Major%20malformations%20associated%20with%20Down,vertebral%20abnormalities.
8. Moller JH, Nakib A, Anderson RC, Edwards JE. Congenital cardiac disease associated with polysplenia. A developmental complex of bilateral "left-sidedness". Circulation. 1967 Nov;36(5):789-99. doi: 10.1161/01.cir.36.5.789. PMID: 6050934.