Arterial Tortuosity Syndrome
a simple twist of fate

"Arterial tortuosity syndrome: early diagnosis and association with venous tortuosity" - Author: Moceri et al. (2013)

Why did they do this study?

- Doctors followed a newborn female who was diagnosed with arterial tortuosity syndrome (ATS) as an infant to better understand the features of ATS as well as the genes responsible for the disease.

How did they do this study?

- Doctors followed the patient from birth to observe her symptoms while performing genetic analysis (taking a closer look at the genes involved in the disease) on the ATS patient.

What did this study find?

- The newborn female was admitted to the intensive care unit because of persistent high blood pressure.
- At this time, doctors diagnosed her with ATS because she had severe twisting of the aorta and pulmonary arteries.
- After her first few months of life, she exhibited the following symptoms:
    - Narrowing of the pulmonary arteries
    - Atypical facial features, such as a long face and a large distance between the eyes
    - Inguinal hernia, when part of the intestine or abdominal wall comes through a point in the abdominal muscle
- Genetic analysis confirmed that the patient had a mutated (altered) SLC2A10 gene. He also exhibited the following ATS symptoms at this time:
    - This gene is a part of the facilitative glucose transporter family, meaning it helps move sugars such as glucose throughout the body.
    - Alterations in this gene are associated with ATS.
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What does this mean for ATS?

- This study provides important information regarding early detection of ATS in newborns as well as the importance of genetic testing to confirm mutations in the SLCA210 gene, which is associated with ATS.