



Arterial Tortuosity Syndrome

a simple twist of fate

“Arterial tortuosity syndrome: clinical and molecular findings in 12 newly identified families” - Author: BL Callewaert et al. (2008)

Why did they do this study?

- Researchers have identified variations in the gene SLC2A10 in patients with arterial tortuosity syndrome (ATS).
- This gene is a part of the facilitative glucose transporter family meaning it helps transport (move) sugars such as glucose throughout the body.
- This study observed newly identified ATS patients with mutations of the gene SLC2A10 to better understand the genetic causes of this disease.

How did they do this study?

- Researchers followed 16 ATS patients and their families.
- All of the ATS patients had the SLC2A10 mutated gene.
- Researchers observed patients' medical conditions and symptoms over time to better understand the progression of ATS.

What did this study find?

- Remarkably, the patients in this study were significantly older than those in previous studies with half of the patients being older than 19.
- All 16 patients had extremely similar clinical characteristics and symptoms including:
 - Twisting of the aorta and larger arteries
 - Abnormal development of the aorta
 - Atypical facial shapes and structures such as a long face, a larger head size, and large ears
 - Cyanosis (a blueish discoloration of the skin due to a lack of oxygen)
 - Difficulty regulating one's body temperature
 - Diaphragmatic hernia, a birth defect in which there is an irregular opening in the diaphragm, the muscle that helps humans breathe
 - Inguinal hernia, when part of the intestine or abdominal wall comes through a point in the abdominal muscle
- Overall, the development of ATS in this study's patients was less severe than previously reported.

What does this mean for ATS?

- ATS symptoms observed in this study were seen in earlier reports.
- However, the study provides encouraging information in that patients were significantly older than those in previous studies.
- Similarly, the overall progress of ATS in these patients was less severe than in other studies, a hopeful and key piece of information for patients and families facing ATS.