"Mutations in the facilitative glucose transporter GLUT10 alter angiogenesis and cause arterial tortuosity syndrome" - Author: PJ Couke at al. (2006)

Why did they do this study?

- Arterial tortuosity syndrome (ATS) is an inherited, recessive genetic disorder that can appear in families that have previously had no members with symptoms.
- ATS is caused by a mutation in a gene called SLC2A10 that creates a protein known as GLUT10. This protein moves sugars such as glucose around the body. People with ATS do not have functioning GLUT10 protein.
- This study explains how this nonfunctioning protein, GLUT10, causes arterial tortuosity syndrome (ATS).

How did they do this study?

- Researchers performed genetic mapping on 6 families with members who had ATS to better understand the complexities of the genes involved in this disease.

What did this study find?

- The presence of a mutation in SLC2A10 was identified as the responsible gene in 4 families with members who have ATS.
- There was no GLUT10 signaling in individuals with ATS in this study.
- People with ATS do not have working GLUT10 protein, which is believed to disrupt the development of connective tissue and the circulatory system.
    - Without working GLUT10, a very important protein called transforming growth factor (TGF-ß) cannot work properly.
        - » TGF-ß plays a critical role in the development of the heart and veins during embryonic development.
    - Patients with ATS do not have normal TGF-ß signaling (it is not known if the signaling is too high or too low).
    - Abnormal TGF-ß signaling has also been linked to other disorders associated with arterial issues including Marfan syndrome and Loeys-Dietz syndrome, highlighting the importance of TGFß in the healthy development of the arteries.

What does this mean for ATS?

- This study suggests that future research should focus on therapeutic ways to intervene with TGF-ß signaling to advance treatment strategies for ATS.