



Arterial Tortuosity Syndrome

a simple twist of fate

“Arterial tortuosity syndrome: phenotypic features and cardiovascular manifestations” - Author: Gayathri Satish et al. (2008)

Why did they do this study?

- This study described the symptoms and characteristics of a 1-year-old, adopted female patient with arterial tortuosity syndrome (ATS).

How did they do this study?

- Doctors followed the young girl and observed her symptoms while also using echocardiographic screening (a type of medical imaging that uses sound waves to create moving pictures of a patient’s heart) to visualize her heart.

What did this study find?

- When the patient was 7 months old, she was referred to doctors because of a recurrent respiratory tract infection (any infection of the sinuses, throat, airways, or lungs).
- At this time, the young patient was diagnosed with ATS because of the following symptoms:
 - Very stretchy and saggy skin
 - Loose joints with an increased range of motion
 - Abnormally long fingers
- During infancy, the patient developed these ATS symptoms:
 - An inguinal hernia, a condition in which part of the abdominal wall or intestines comes through a point in the abdominal muscles
 - An umbilical hernia (when part of the intestines, fat, or fluid pushes through the abdominal muscles near the belly button)
 - Downward slanting of the eyes
- Echocardiogram imaging showed the following characteristics typical of ATS:
 - A hugely enlarged ascending aorta
 - Twisting of the aorta
 - An enlarged pulmonary artery
 - Narrowing of the small segment of the thoracic aorta

What does this mean for ATS?

- This study provides important information regarding the symptoms and diagnosis of ATS, which are consistent with other studies on the disease.