



## “Cardiovascular findings in arterial tortuosity syndrome” - Author: R Hoop et al. (2006)

### Why did they do this study?

- This study followed a young male patient with ATS to point to the importance of early medical imaging and screening to differentiate between different connective tissue disorders and to detect symptoms of ATS early on.

### How did they do this study?

- This study used echocardiographic screening (a type of medical imaging that uses sound waves to create moving pictures of a patient's heart) to visualize the heart of a young male with ATS.

### What did this study find?

- At birth, the infant experienced the following ATS symptoms:
  - A diaphragmatic hernia, a birth defect in which there is an irregular opening in the diaphragm, the muscle that helps humans breathe
  - 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
  - Overall muscle weakness
- The magnetic resonance (MR) imaging (an imaging technique to examine the anatomy and function of the body) showed the following ATS symptoms:
  - The lengthening and twisting of the aorta
  - Severe twisting of the pulmonary arteries
- When visualizing the coronary arteries, both arteries had a twisting path without any abnormal widening or narrowing.

### What does this mean for ATS?

- ATS symptoms in this study were typical of ATS characteristics seen in other studies.
- The study explains how the earlier echocardiographic screening is used, the better influence it can have on the detection, prognosis, and treatment of ATS.