# "Cardiovascular findings in a boy with arterial tortuosity syndrome: case report and review of the literature" - Author: Filiz Ekici et al. (2011)

## Why did they do this study?

- This study describes the symptoms and characteristics of a 5-year-old boy with arterial tortuosity syndrome (ATS).

## How did they do this study?

- Researchers observed symptoms of the patient and used vascular imaging (pictures of vessels) to visualize cardiac abnormalities in the patient.

## What did this study find?

- When the patient was 1-year old, he was initially referred to the pediatric cardiology department because of the following symptoms:At this time, the young patient was diagnosed with ATS because of the following symptoms:
    - Heart murmur (irregular heart sounds)
    - Shortness of breath
    - Fatigue (tiredness)
    - Cyanosis (lack of oxygen, resulting in a bluish color of the skin)
- When he turned 1 year old, he underwent an operation for a right-sided inguinal hernia (when part of the abdominal wall or intestines comes through a point in the abdominal muscles).
- He also exhibited the following ATS symptoms at this time:
    - Soft and very stretchy skin
    - Atypical facial features, including a down slanting face, beaked nose, large upper lip, and skin lesions (cuts)
- Magnetic resonance imaging (a type of medical imaging to better see the anatomy and function of different body parts) showed the following symptoms:
    - Twisting of the aortic arch
    - Twisting, elongation, and right-sided kinking of the thoracic aorta
- At 8 years old, he had another operation for a right-sided inguinal hernia.
- At the end of the study (2011), the patient was alive and 10 years old.
    - At this time, he suffered from recurrent chest pains from physical activity.

## What does this mean for ATS?

- This study provides important information regarding the symptoms of ATS in young patients.
- The researchers also recommend a closer examination of patients' symptoms, including an analysis of the skin, molecular studies of the genes involved, and quantification of patients' plasma levels, to help differentiate among different types of connective tissue disorders.