Cardiovascular Features in Cutis Laxa

Suneeta Madan-Khetarpal
Cutis Laxa Clinic
Children’s Hospital of Pittsburgh of UPMC
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Not every person with cutis laxa will have cardiac involvement.

Certain forms of cutis laxa have specific cardiac symptoms.

Symptoms can range from aortic dilation and aneurysm, supravalvular aortic stenosis, arterial stenosis, and arterial tortuosity.

Echocardiograms (ECHOs) can be done to detect some of these cardiac conditions.
Aortic Dilation (Aneurysm)

- Aortic dilation is the swelling / ballooning of the aorta, which is the largest artery in the body.

- This can happen if the elastic fibers in the aorta cause it to become stretched.

- Forms of cutis laxa that may involve aortic dilation are:
  - FBLN4 (recessive type 1A)
  - ELN (dominant type)
  - Acquired form

http://en.wikipedia.org/wiki/Aorta
Dominant Cutis Laxa (ELN)
Aortic aneurysm and tortuosity in ARCL1 caused by a fibulin-4 mutation

Aortic root aneurysm
Arterial (or Vascular) Tortuosity

- Arterial tortuosity is the lengthening and twisting of arteries in the body, usually the larger arteries, including the aorta.

- Sometimes this is seen in people who have recessive cutis laxa type 1A (FBLN4).

Supravalvular Aortic Stenosis (SVAS)

- Aortic valve stenosis is when the opening for the aortic valve in the left ventricle gets more narrow than expected.
- This can be seen in people who have recessive type 1B (FBLN5) and dominant type (ELN).
Regurgitation

- Regurgitation occurs when the blood moves in the opposite direction, which can occur if there is weakness in the valves.
- In aortic regurgitation, this can cause the left ventricle to become dilated, making it more difficult for the heart to pump blood through the aorta, which leads to the rest of the body.
- Sometimes this is seen in people with dominant cutis laxa (ELN).
Pulmonary artery stenosis is then narrowing of the pulmonary artery.

- This can cause shortness of breath, fatigue, and a fast heart rate.
- This can be found in some people with URDS cutis laxa (Urban-Rifkin-Davis Syndrome) which is in the gene LTBP4.

[Image showing pulmonary artery stenosis and associated lung structures]