Why the subject HTAAD?

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What I knew about HTAAD in 2008-2010

- Marfan syndrome: A connective tissue disorder with (thoracic) aortic dilation and dissection
- Loeys-Dietz syndrom: A new, rare, aggressive Marfan-like condition. 2 subtypes, not defined by the 2 genes
- Vascular EDS: A rare subtype of EDS with arterial complications
- No matter what happens, there will be too much TGFB signalling

The development since then

- OMIM: Loeys-Dietz syndrome subtypes 1-5, defined by genes
- An increasing number of known genes that gives HTAAD, currently > 30 (Bradley 2016)
- Syndromic vs non-syndromic HTAADs – maybe not so clearcut?
- More emphasis on genes, not just diagnostic criteria – consequences?

Current simplified understanding of HTAAD

- Changes in fibrils in the extracellular matrix
- Disturbances in the TGFB pathways
- Changes in the contractile apparatus inside vascular smooth muscle cells

- Everything seems to be connected
- My guess: still a vast number of genes waiting to get unveiled

Norway 2016

- Gene panel with HTAAD genes
- Testing of new patients with HTAAD and patients with eg genetically unconfirmed «Marfan syndrome»
- **TRS has formal competence center responsibility for LDS**
- **HTAAD is a new group at TRS this year**

Current challenges

- We meet patients with diagnoses that we have never seen before
- Patients turn out to have another diagnosis than we previously thought
- There is a lot of new knowledge
- Our time is limited, hard to keep up
- Sometimes difficult to understand the articles
- Clinical findings may differ from our expectations

Solutions

- Collaboration - between different specialists and over borders!
- Learning from each other and exchanging our experiences
- Our systems have different strengths and weaknesses – let's take advantage of that
- In Norway: patients with HTAAD should be referred to TRS so we can meet them and learn from them