FBN1 mutation in our family.
Personal history
DNA discovery
Distribution of the mutation.
Reactions and Concerns.
Personal Medical History

- 09/1937 born in Maastricht, Holland. (78 now)
- 1995 Abdominal Aortic Aneurysm repair.
- 2006 My sister suffers a dissection
- 2007 Left iliac aneurysm embolised.
- 04/2008 Right popliteal aneurysm repair
- 06/2008 Left popliteal aneurysm repair
- 01/2012 Right femoral aneurysm repair
- 10/2013 Left femoral aneurysm repair.
- 01/2014 Right popliteal repaired with Viaban Stent.
- 03/2014 DNA test
- 01/2015 Aortic root and valve repaired Bental's procedure + Pacemaker.
- 10/2015 Acute thrombosis of Viaban Stent.
- On going surveillance of juxtarenal aneurysm
New FBN1 pathogenic variant.
From the report of the Collagen Diagnostic lab, University of Washington. (March 2014).

- No mutation in COL3A1, TGFBr1, TGFBR2, ACTA2, SMAD3, TGB2. (The usual suspects)
- Identification of a likely pathogenic variant in one allele of the FBN1 Gene, that encodes the protein fibrillin 1.
- The variant is predicted to result in substitution of arginine (Arg) by Cystine (Cys) at protein position 2360.
- Not reported in UMD-FBN1 mutations database, dbSNP, ClinVar or Exome Variant Server.

Taken altogether, the available information supports that this is a likely pathogenic variant that explains the vascular phenotype in the patient.
FBN1 mutation in our extended family
2014
FBN1 mutation in our extended family
2016
Summary
(excluding descendants of Ae4)

- Of my father's 36 descendants, 15 have been identified as carrying the mutation. (42%)
- If one carries the mutation one is highly likely to be tall, but one can be tall without the mutation. (4 cases)
- Already 6 members have been operated for enlarged aortas: M (77), R (71), C(61), X(52), B (44), R (24).
- Enlarged aortic root is emerging as a common feature.
- Although there is a small sprinkling of Marfan features there is no consistency or pattern.