Congenital Aortopathies
Marfans, Loeys-Dietz, ACTA 2, etc.

DATE: October 9th, 2017 PRESENTED BY: Cristina Fuss, MD
24 yoF present with SoB
24yo F

- Presenting to local ED with SoB
- No other pertinent history at that time available
- Symptomatic 5 cm ascending aortic aneurysm, coarctation
- Bentall procedure with mechanical aortic valve, hemiarch replacement and coarctation repair
- Referral to OHSU aortopathy center
Thoracic Aortic Aneurysm/Dissection

• Rising incidence
• Syndromic vs. non-syndromic
• Requires understanding of family history
• Render proper diagnosis which steers treatment
Thoracic Aortic Aneurysm/Dissection (TAAD)

- Occurs sporadic
- Familial clustering in > 20% of cases
- Positive family history increases RR by 6-20 fold
- Usually autosomal dominant with variable expressivity
Familial TAAD

- Younger age at onset
- More aggressive pathology
- Often with bi-commissural aortic valve
24yF

- Additional history
  - Surgery at age 4 mon for
    - PDA
    - Pulmonary artery aneurysm
ACTA 2 Features and Gene loci

• ACTA 2 gene
  – Smooth muscle alpha-2 actin
  – Associated with
    • PDA
    • Aneurysms (aorta and others)
    • Moyamoya
    • Bladder dysfunction
    • Digestive tract dysfunction
Familial TAAD

- Marfan Syndrome
- Loeys Dietz Syndrome
- Vascular Ehlers Danlos Syndrome
- Bicuspid aortic valve
- ACTA 2
Marfan Syndrome

- First described in 1896 by Dr. Antoine Marfan
- Mutation in the FBN1 gene (chromosome 15)
- Increased production of TGF-beta
- 75% inherited, 25% new mutations
- Autosomal dominant
Marfan features

- Aortic aneurysms
- Mitral valve prolapse
- Pneumothorax
- Scoliosis
- Pectus deformity

- Arachnodactyly
- Severe nearsightedness
- Pectus deformity
- Flexible joints
- Flat feet
- Crowded teeth
- Stretch marks unrelated to weight gain/loss
- Ectopia lentis
- Detached retina
- Early glaucoma and cataracts
Marfan Syndrome Diagnosis

- Revised Ghent criteria
- Clinical and genetic
  - 2 clinical
  - or
  - 1 clinical + 1 genetic criteria
- Clinical score system; max 20 points
  - ≥ 7 points – systemic involvement
Loeys-Dietz Syndrome

- Types I-V
  - Depending on their genetic cause
    - TGFBR1
    - TGFBR2
    - SMAD3
    - TGFBR3
  - Autosomal dominant with 75% new gene mutations
- Variable severity
- Manifest anytime between childhood or adulthood
Loeys-Dietz Syndrome

• Characteristics:
  – Aortic aneurysm
  – Dissection
  – Arterial tortuosity
  – Cranial synostoses
  – Scoliosis
  – Pectus deformity
  – Joint contracture/club foot
47yM LDS routine screening

October 2014

January 2012
24yoF

- Re-established care 20 weeks pregnant
- High risk pregnancy
- Oligohydramnios
- Multiple fetal/neonatal abnormalities
  - Patulous bladder
  - Abdominal wall distension
  - Dilated PA and ductus
  - Congenital mydriasis
  - Malrotation
ACTA2 mutation

- ACTA2 gene:
  - Smooth muscle alpha-2 actin
- Familial thoracic aneurysm and dissection
  - >30 gene mutations
- Associated disorders:
  - Impairment of smooth muscle function
    - Bladder
    - Digestive tract
    - Moyamoya disease
    - Premature CAD
ACTA2 mutation features

- **TAAD**
  - Type A dissection (54% at median 27 years)
  - Type B dissection (21% at median 36 years)
  - Early dissection (< 5cm) peripartum!

- **Other:**
  - Occlusive vascular disease (CAD, Moyamoya)
  - PDA
  - Iris flocculi
  - Livedo reticularis
ACTA2 follow up
Imaging

• Regular screening of young patients
• MRA
  – Loeys Dietz protocol
    • MRA CAP
    • MRA neck
    • MRA COW
• CTA
  – Chest (gated!)
  – Abdomen/pelvis
Aortic measurements

- Cross-sectional oblique
- Standard levels (list!)
  - Sinus of Valsalva (root)
  - ST junction
  - Ascending Ao (level of RPA)
  - Arch
  - Descending Ao (level of RPA)
  - Hiatus
  - Level of SMA
  - Level of RA
  - Bifurcation
- Don’t forget visceral and pelvic vasculature
Definition

- Ectasia
  - < 150% of normal
- Aneurysm
  - > 50% increase in diameter compared to normal
- Z-score
  - Standard deviations above/below age-specific mean
    - \( Z = \frac{(x - \mu)}{\sigma} \)
    - \( x \): observed measurement, \( \mu \): population mean, \( \sigma \): population standard deviation
Recommendation for Surgery

• Sporadic
  – $\geq 55$ mm
  – No BaV
  – Increase in diameter $\leq 5$ mm/year
Recommendation for Surgery

- Familial
  - $\geq 50 \text{ mm (MFS)}$
  - Or
  - $\geq 45 \text{ mm (with risk factors)}$
    - Family history of dissection
    - Rapid increase in size
    - Sever aortic/mitral regurgitation
    - Pregnancy
9 yF LD
Thank You