The Interventional Radiologist's Role in the Treatment of Tissue Overgrowth Disorders which are associated with Vascular Malformations

The Emory University Medical Center Experience
Atlanta, Georgia USA

I have no conflict of interest to declare

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Goal of this presentation
- To review the embryogenesis of the vascular malformations encountered in the Overgrowth syndromes
- To review the Emory experience treating Tissue Overgrowth Syndromes that are associated with Vascular Malformations
- To encourage you to incorporate this patient group into your IR practice

Embryogenesis of venous malformations
- The clinical behavior of the malformation depends on the embryogenic stage at which it occurs
- Vessels remain in the form of reticular networks when development is early. These proliferate upon traumatic or hormonal stimulation
- When defective vascular development occurs in a later stage they involve named vessels such as the Popliteal v., Femoral v., and IVC.
- These embryologically mature veins no longer possess the capacity to proliferate
- These are called Extra-truncal and Truncal by the Hamburg Classification

Association of port wine stains and Low Flow shunts in Overgrowth Syndromes
- KTS
- CLOVES syndrome
- Maffucci syndrome
- Blue Rubber Bleb Naevus syndrome

Association of port wine stains and High Flow shunts in Overgrowth Syndromes
- Parkes Weber syndrome
- CV-AVM (capillary malformation-arteriovenous malformation syndrome)
- Cobb syndrome
- CLOVES syndrome
- Blue rubber bleb nevus syndrome (Bean syndrome)
- Bannayan-Riley-Ruvalcaba syndrome
- Proteus syndrome
The IR Toolbox

- Sclerotherapy
- Cutaneous laser treatments
- Compression garments

Compression hose are an important element in the treatment of overgrowth syndromes

History of KTS

- First described by French physicians Maurice Klippel and Paul Trénaunay in 1900 as “Naevus Vasculosus Osteohypertrophicus”
- Frederick Parkes Weber described similar cases in 1907 and 1918, which additionally had high flow AVMs

3 Main Features of KTS

1. Capillary malformation “Nevus flammeus” (port-wine stain)
   - One or more with very sharp borders
   - Typically on the lateral aspect of the affected limb in association with the lateral marginal vein
   - May be absent in very rare cases “atypical KTS”
2. Venous and lymphatic malformations
   - (macro and micro cystic)
3. Soft tissue, fatty and bony overgrowth of a limb
### Components of the KTS venous malformation

- Immature reticular veins (Extra-truncal veins)
- Truncal venous malformations that present as a persistent fetal remnant veins that have failed to involute and regress normally.
  - Marginal and Lateral embryonic veins and the Sciatic vein
  - A prominent marginal venous system and a deep venous system typically coexist in KTS.
- Marginal veins cannot be removed or occluded before determining the patency of the deep venous system
- Also frequently have pelvic phlebectasia and

### Important missing features of KTS

- Not associated with high flow AVMs
- Not associated with the GI and GU systems
- Not associated with CNS
- Not associated with spinal disorders

### KTS Genetics

- Sexes affected equally
- Affects all racial groups
- Not certain whether or not it is genetic in nature
  - There is some evidence that it may be associated with a translocation at t(8;14)(q22.3;q13)
  - Some researchers have suggested a VG5Q association.

### Surgical treatment of KTS

- Mayo Clinic has the largest reported experience
- 252 cases in 39 years
  - 58% treated surgically
    - 40% varicose veins
    - 60% venous malformations
    - Debunking operations in 65%
  - High recurrence rate in all operations
    - (>50%)

### KTS - Laser treatment of weeping lymphatic vesicles

- 10 year old male with KTS
- Treated many times for RLE pain and swelling
- Pain in right leg now interfering with his school work and other activities.

### KTS - sclerotherapy

- 10 year old male with KTS
- Treated many times for RLE pain and swelling
- Pain in right leg now interfering with his school work and other activities.
Painful areas marked pre-sclerosis

MRI performed in 2010

STS-foam sclerotherapy 10-31-2013

Parkes Weber syndrome

- Classified as Parkes Weber syndrome if an AVM is present
- Characterized by faint capillary stains on an overgrown limb
- Unlike KTS, the phlebectasia is due to high flow and not dysplasia.
- Has diffuse hypervascularity and a risk of cardiac overload

- Not a type of KTS with AVM, therefore the triple eponym Klippel Trenaunay Weber syndrome (KTWS) should be abandoned

33 year old male with Parkes Weber Syndrome

- Complains of increasing pain in the left leg

Pelvic venous congestion and large venous aneurysm compressing the bladder
IVC and left iliac venomegaly

Increased arterial flow to the LLE

Popliteal and ATA aneurysms, PA and PTA OK

PTA aneurysm eroding the tibia

Embolized the ATA one month later

More work to be done
CLOVES syndrome

- Congenital
- Lipomatous
- Overgrowth
- High flow vascular malformations
- Epidermal nevi
- Spinal/Scoliosis/Seizures/Skeletal anomalies

**Peraspinal AVMs** are common and associated with significant morbidity.

- One of the common vascular findings in this syndrome is a paraspinal-truncal arteriovenous shunt associated with a port wine stain and truncal overgrowth
- Associated with phlebectasia and increased incidence of pulmonary embolism

16 year old female with CLOVES

**Painful mass above left breast**

**S/P forearm amputation**

Needle position on T2, sclerosant/contrast injection on T1

Proteus syndrome

- Rare mosaic progressive sporadic disorder with a spectrum of clinical features, including:
  - Connective tissue nevus
  - Epidermal nevus
  - Disproportionate progressive overgrowth
  - Tumors
  - Slow-flow vascular malformations are reported to be common
- AVMs are uncommon
- Identified an activating mutation in AKT1 kinase in a mosaic state gene in 26 of 29 patients with the syndrome
Thank you!