Clinical Institute
Humanitas “Mater Domini”
Castellanza (Varese) Italy

Center for Vascular Malformations
“Stefan Belov”
Founded in 1992
Combined malformation: Klippel-Trenaunay syndrome
Conflict of interest

I have no conflict of interest
Arguments to be discussed

1. What is Klippel Trenaunay
2. What is NOT Klippel-Trenaunay
3. What kind of anomalies exist in KTS
4. How to manage
What is KTS?

• Classical definition:

KTS is a complex vascular malformation with the typical “triade” of signs:

- nevus
- dilated superficial veins
- limb overgrowth

A pure clinical definition
Maurice Klippel and Paul Trenaunay (1900)

- Described cases with varicose veins, limb hypertrophy and cutaneous nevi: the «triade», Klippel-Trenaunay Syndrome (KTS)
- No signs of arterial involvement (NO a-v defects)

No diagnostic tools!
Frederick Parkes Weber (1907, 1908, 1918)

- Described cases of “Hemangiectatic hypertrophy of limbs”
- These cases are similar to K-T (with the “triade”) but with a-v fistulas
- They are called «Parkes-Weber Syndrome» (PWS)
Which is the definition of Klippel–Trenaunay syndrome?

- “A mesodermal abnormality that involves veins, capillaries, soft tissue and skeletal elements”
  
  Pearce, Yao et al, 1991

- “A capillary-lymphatic-venous malformation associated with soft tissue/skeletal hypertrophy, usually to one or more limbs and sometimes the trunk”
  
  Connors, Mulliken, 2005

- “When a-v malformations coexist, it is called “Klippel-Trenaunay-Weber”
  
  Bothun et al, 2011
More definitions

• “Klippel-Trenaunay-Weber… cutaneous hemangiomas, venous varicosities, and osseous-soft tissue hypertrophy of the affected limb…. without a-v fistulas”

**Boutarbuch, 2010**

“K-T is a complex malformation with the triad and occasionally limb hypotrophy and including lymphatic components”

**Gloviczki, 2007**
“Ectopic” K-T syndromes in the literature

- Ear a-v Managa, 1980
- Pelvic a-v maformations Groneweg, 1987
- Gingival hyperplasia, «hemangiomas» of thongue, lips and gums Ozdemir, 2010
• The “Klippel-Trenaunay-Parkes-Weber” eponym is meaningless and should be abandoned......
Vascular defects with the “triade”

- Aplasia of deep veins
- Hypoplasia of deep veins
- Truncular superficial dysplastic veins with or without marginal vein
- Extratruncular areas of dysplastic veins also with truncular
- Truncular venous + extratruncular lymphatic dysplasias
- Extratruncular venous + extratruncular lymphatic dysplasias
- Extratruncular venous + truncular lymphatic dysplasias
- Truncular venous + AVM: PWS
- Extratruncular venous + AVM: PWS
- Truncular lymphatic + AVM

The “triade” is not a criteria to define KTS!
What is a syndrome?

• The term derives from the greek “sundrome”, that means “concurrence of symptoms, concourse”

• Referred to association of several signs, symptoms or phenomena that often occur together

• Regarding CVM it regards only a combination of vascular defects and NOT a single one
Cases to be defined as KTS

- Diffuse VM on the whole limb
- Combination of at least 2 CVM
- Venous malformation should be present
- Truncular and extratruncular VM combination meet the definition
- Lymphatic malformation, truncular or extratruncular, can be present but is not mandatory if two VM coexist

Lee BB et al, VM consensus, Int Angiol, 2013
What is not KTS

• Cases with only nevus
• Cases with AVM
• Cases without venous anomalies
  (involving not the whole limb)
• Cases with malformations in other body areas (face, trunk, abdomen)
No KTS

Pure capillary malformation

AVM

Lymphedema

AVM+Lymphatic dysplasia
No KTS:
Truncular + extratruncular LM

Extratruncular areas of dysplastic lymphatics

Aplasia of main lymphatics
No KTS

Venous malformations limited on a segment of the limb

Venous malformations sited in other parts of the body
How to approach KTS?

• Avoid diagnosis without tests
• Perform a step by step diagnosis
• Proceed from the less invasive to the more invasive procedure

Data of each step should guide to the next
Clinical data

No nevus

Limb lengthening

Limb shortening

Bilateral
Foot hypertrophy
Clinical data noticed
(46 cases, Castellanza 2011 – 2015)

- Right limb affected .............. 18 (39%)
- Left limb affected ............... 18 (39%)
- Bilateral disease .................. 10 (22%)
- Nevus ................................ 42 (91%)
- Dilated superficial veins ....... 46 (100%)
- Limb overgrowth ................ 18 (39%)
- Limb shortening ................. 3  (7%)
- No limb length difference ...... 25  (54%)
- Foot overgrowth ................. 4  (9%)
Diagnostic steps

1) clinical examination
2) Duplex scan
3) Plain Rx of the limbs
4) MR or CT
5) Lymphoscintigraphy
6) Other test to complete diagnosis
Nevi, dilated superficial veins and limb overgrowth

Marginal vein

Lymphatic defects
2) **Duplex scan**

- **Exclude AVM**
- **Study main veins anomalies** (aplasia, stenosis, dilatations)
- **Study infiltration of tissues**
- **Find out type of flow inside abnormal vascular areas** (high flow, low flow, no flow)
High flow = AVM
Low flow = VM
No flow = LM

Venous hypoplasia
Venous aplasia
Venous + lymphatic
Phlebolyth
Venous aneurysm
Marginal vein with reflux

Venous
Lymphatic
3) Plain Rx exam
Aplasia of left iliac vein
Aplasia of sup. femoral vein
Infiltrating VM
Marginal vein
Lymphoscintigraphy

- Demonstrate truncular lymphatic defects
- Should be performed with separate study of deep and superficial lymphatics
Vascular defects recognized
(46 cases, Castellanza 2011-2015)

- Deep vein aplasia ............... 9 (19%)
- Deep vein hypoplasia .......... 9 (19%)
- Deep infiltrating veins .......... 19 (41%)
- Marginal vein .................... 14 (30%)
- Sciatic vein .......................... 3 (7%)
- Superficial dysplastic veins .... 46 (100%)
- Truncular lymphatic defects ... 13 (28%)
- Extratruncular lymphatics ...... 2 (5%)
Treatment possibilities

- Surgery
- Foam sclerosis
- Percutaneous alcohol treatment
- Interstitial laser treatment
Conclusions

• **KTS is a complex CVM involving the whole limb with possible extension**

• **Several type of vascular defects are possible, also in combination**

• **Diffuse venous dysplasia is the main defect**

• **Combination with lymphatic defect is possible but not mandatory**

• **Limb overgrowth exist in 50% of cases**

• **Treatment is possible if a correct diagnosis is performed**
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