Vascular Anomalies: Diagnosis and Management
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Disclosure
In the past 12 months, I have no relevant financial relationships with the manufacturers of any commercial products or providers of commercial services discussed in this CME activity.

What is this lesion?

Learning Objectives
• Define "vascular anomalies"
• Learn approach to classification
• Review diagnosis and management of specific lesions
  – Alimentary tract (exclude hepatic lesions)

Definition of vascular anomalies
• Morphologically and biologically diverse group of abnormalities of vascular channels
• Often congenital but grow or change over time
• May involve single or a combination of vessel types: capillary, venous, arterial, and lymphatic
• May represent isolated lesions or part of multiorgan system condition

Classification
• Primary classification
  – Tumors
  – Malformations
• Based on biological behavior, clinical behavior, and physical endothelial characteristics
• Predicts likely response to antiproliferative drugs vs surgical or other ablative therapy

**Classification for Vascular Anomalies**

International Society for the Study of Vascular Anomalies (ISSVA)  
(Approved at the 20th ISSVA Workshop, Melbourne, April 2014)

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<th>Vascular anomalies</th>
<th>Vascular malformations</th>
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* defined as two or more vascular malformations found in one lesion

**Clinical GI Presentations**

- Gastrointestinal bleeding  
  - Overt  
  - Occult with chronic anemia
- Abdominal pain, intestinal obstruction
- Intestinal malabsorption, PLE, diarrhea, hypoalbuminemia, ascites

**Hemangioma**

- Most common tumor of infancy and childhood
- Incidence (neonates) 1-2.6%  
  - <1 kg premi 23%  
  - <1.5 kg 15%
- Gender = 3:1 F:M
- Distribution  
  - Solitary 80%  
  - Multifocal 20%  
  - Often with intracranial or visceral involvement
- Three growth phases  
  - Proliferative neonate  
  - Involution (early) 1-5 y  
  - Involution (late) >5 y

**Intestinal Hemangioma**

- 16 children in BCH VAC database 1998-2012
- 14F: 2M  
  - 14 (88%) presented with GI bleeding in first 4 mos of life  
  - 2 bowel perforations

Soulkoulis et al (manuscript submitted)

**Hemangioma**

- Diagnosis (n)  
  - Laparotomy 8  
  - CT/MRI/US 5  
  - Angiogram 1  
  - Colonoscopy 1  
  - WCE 1  
  - EGD/colon (-) 8/9
- Involved site  
  - SB 15  
  - Colon 1  
  - Cutaneous 9 (56%)

CT with enhancing bowel wall thickening
Intestinal Hemangioma

- 7/16 with congenital abnormalities
  - Pierre Robin sequence
  - Short bowel and malrot
  - VSD and pulm HTN
  - Tetralogy of Fallot
  - Cleft lip/palate, micrognathia
  - Tailgut cyst
  - PHACES association (n=1)
    - Posterior fossa tumor
    - Hemangioma of face
    - Arterial abnormality
    - Cardiac anomaly
    - Eye abnormality
    - Sternal defects

Intestinal Hemangioma: Treatment

- Therapeutic options
  - Propranolol (2 mg/kg/day)
    - Should be first choice
  - Corticosteroids (2-4 mg/kg/day)
    - Well established efficacy
  - Vincristine (low dose)
  - Combination drug therapy
  - Bowel resection (last resort)

- Duration of therapy
  - Usually 6-9 months, but sometimes longer
  - Major endpoint is cessation of bleeding

Blue Rubber Bleb Nevus Syndrome (BRBNS)

- Multifocal polyloid VM
  - “cavernous hemangioma”
- Incidence unknown
- Somatic mutation in TIE2 gene, chromosome 9p21.2
- Predominant skin and intestine involvement
- Presents with GI bleeding in early childhood
- Diagnosis by endoscopy

Beware of endoscopic mimicry!

BRBNS

- Soft tissue lesions detected by labeled RBC scan
- Intussusception of small bowel may be asymptomatic or present with intermittent pain or obstruction

BRBNS: Treatment

- Surgical excision is preferred definitive therapy
  - May be supplemented by endoscopic ligation/polypectomy
- Limited role for antiproliferative drugs
  - Delay surgical intervention in young children
  - Rescue therapy for poor surgical candidates
  - Sirolimus is current drug of choice

**Reticular Venous Malformation**

- Superficial VM with a tortuous variceal or net-like pattern
- Often involves entire length of colon and distal ileum
- May include other SB
- Incidence is unknown
- Usually sporadic, rarely hereditary
  - Autosomal dominant familial colonic varices
  - Turner’s syndrome

**Reticular VM: Treatment**

- Reduce severity of bleeding episodes with antibrinolytic or clot stabilizing drugs, eg. tranexamic acid (Lysteda) or aminocaproic acid (Amicar)
- Endoscopic sclerotherapy or thermal coagulation/ablation, eg. argon plasma coagulation
- Colectomy or segmental bowel resection

**APC ablation**

- Start VID 003 at 08:17

**Segmental Colonic VM**

- Endoscopic appearance
  - Diffuse erythema, flat or slightly raised texture, absent branching vessel pattern
- CT or MRI shows wall thickening due to transmural infiltration
- Frequent rectosigmoid distribution
- Isolated lesion, or with Klippel-Trénaunay Syndrome

**Segmental Rectosigmoid VM: Therapy**

- Transanal sclerotherapy
- Partial colectomy with pull-through anastomosis

**Segmental Lobular VM**

- Raised lobular surface
- Significant mass effect
- Focal
- Transmural
- Sporadic
- Treated by surgical resection

What anatomy is involved?

CUTANEOUS ANGIOMATOSIS WITH THROMBOCYTOPENIA (CAT)

- Also called 'multifocal lymphangioendotheliomatosis with thrombocytopenia (MLT)*'
- Multiple red, purple, or brown cutaneous lesions
- Hemangioma-like gastrointestinal and pulmonary lesions
  - Also liver, spleen, muscle, bone, brain, retina
- Treat with corticosteroids, vincristine, thalidomide to control bleeding
- Partial regression over months to years but long-term outcome unknown

*North et al, Arch Dermatol 2004;140:599

PROVISIONALLY UNCLASSIFIED VASCULAR ANOMALIES

Summary

- Careful gross anatomic, endoscopic, and microscopic description of GI vascular anomalies is essential for correct classification
- Classification and distribution of lesion(s) determines most effective treatment
- A coordinated multidisciplinary approach facilitates optimal management and advances scientific understanding

INTESTINAL LYMPHANGIECTASIA

- Predominantly small bowel involvement
- Essential role of wireless capsule endoscopy
- White-tipped thick villi
- Patchy multifocal or continuous
- Malabsorption
  - Protein-losing enteropathy
  - Fat malabsorption
- Treated by reduced long chain fats, MCT oil, albumin infusions, octreotide, bowel resection
- Potential role for lymphangiography to look for disrupted thoracic duct
  - Microsurgical reconstruction?

Valuable endoscopic Mapping!

Distal stomach
Duodenum