KOMPRESNÍ SYNDROMY GIT A EHLER DANLOS A JEJICH VÝZNAM

PETR WOHL

PROČ TOTO TÉMA ?

1. diagnostická oblast - diagnóze se často nevěří – psychiatrie….

2. léčebná: obavy a nejistota z výkonů

3. určité množství nemocných (**celosvětově narůstajících**) končí v ambulancích pro DPV či intestinálního selhání (data z UK)

4. určité množství nemocných je pak **indikováno k DPV**…..

A málokdo se tím zabývá systematicky…….

5. Zásadní roli - **zobrazovací metody**…….a schopnost rozeznat vztah: nález – příznak…….

ÚVOD

- Vaskulární abdominální (a pelvické) kompresní syndromy (Wilkieho syndrom, Dunbarův syndrom, Nutcracker syndrom, May-Thurnerův syndrom atd.) jsou relativně vzácné a často nediagnostikované pro zcela nespecifické potíže.

- Oligosymptomatické formy zůstávají dlouho nediagnostikované, ale v případě plné manifestace, jsou velmi závažné a mohou ohrozit nemocné i na životě.

- V praxi je mnoho situací, kdy se diagnóza zcela zpochybňuje a nemocní často dostávají psychiatrickou diagnózu, nebo jsou zařazení do funkčních poruch GIT.

MOŽNÉ KOMPRESE.....

- Fraley's **syndrome** is a rare cause of obstruction of the collecting system. It results from an extrinsic **vascular compression** of a calyceal infundibulum by a branch of the renal artery
- Pinch-off **syndrome** (POS) occurs when a long-term central venous catheter is **compressed** between the clavicle and the first rib.
- **Ovarian vein syndrome** corresponds to ureteral compression by a dilated ovarian vein.
- Popliteal artery entrapment **syndrome** (PAES) is a rare congenital **vascular** pathology caused by the **compression** of the popliteal artery by adjacent muscle and tendinous structures
- **Vascular compression** of the vestibulocochlear (VIIIth) nerve may cause constant or recurrent positional vertigo, tinnitus, and/or hearing loss.

MOŽNÉ KOMPRESE.....

- Reversible gastroparesis: functional documentation of celiac axis compression syndrome and postoperative improvement (2006 Am. Sur.) – **symptomy gastroparézy i mezenteriální ischemie**
- Mediastinal goiter as a cause of superior vena cava **syndrome** and tracheal **compression**
- Iliac vein compression syndrome is the phenomenon of nonthrombotic iliac vein obstruction caused by compression of left iliac vein between the right iliac artery and fifth lumbar vertebra.

- **Kompresí je dost……**
- We have observed a tendency to claim a psychosomatic explanation for the symptoms of CS in many patients with hEDS and similar disorders without even considering CS. Am J Med Genet. 2021;1–9.

Median arcuate ligament syndrome	Young females, post-prandial pain, nausea, and weight loss.	MAL thickness > 4 mm. "Fish hook" or "J"-shaped celiac artery impression. Post-stenotic dilation of celiac artery Collaterals between the SMA and celiac artery. True compression persists in the end-inspiratory phase.	Celiac artery decompression. Young patient ideal for surgery.
Superior mesenteric artery syndrome	Recent history of marked weight loss. Relief of obstruction in prone or left lateral decubitus position.	AMA < 22° (normal 28–65°) and an AMD < 8 mm (normal 10–34 mm). Vertical extrinsic impression on the third portion of the duodenum.	Acute: nasogastric tube decompression, nasojejunal feeding. Chronic: open or laparoscopic duodenojejunostomy
Nutcracker syndrome	Microscopic to severe hematuria, orthostatic proteinuria, left flank pain. Left varicocele in males and left pelvic and vulvar varices in females. Bimodal peak (see text), thin/asthenic body habitus.	Abrupt narrowing of the LRV between the SMA and aorta "beak sign." Ratio of LRV diameter at the renal hilum and at aortomesenteric region of more than 4.9. LRV to inferior vena cava gradient of more than 3 mmHg in invasive venography diagnostic.	Conservative: young, mild symptoms. Surgery: LRV bypass, transposition to inferior IVC, venous stenting, renal auto transplantation, and left nephrectomy.
May-Thurner syndrome	Young and middle-aged females. Unilateral left lower limb DVT.	Left CIV compression by the right CIA. Left CIV thrombus and/or tortuous venous collaterals.	Relieve mechanical compression with endovascular stent.
Uretropelvic junction obstruction by crossing renal vessels	Renal colic, hematuria, UTI. Dietl's crisis.	Anterior crossing renal vessels, especially the lower pole segmental vessels. Vessels within 1–2 cm from UPJ. Late arterial phase ideal to detect branch from renal artery/vein of accessory branch from the aorta, IVC, or iliac vessels.	Laparoscopic dismembered pyeloplasty.
Retrocaval ureter	Asymptomatic, flank pain, hematuria, UTI. 3rd or 4th decade of life due to hydronephrosis.	"J" shape, "Fish hook," or "S"-shaped proximal hydroureter and resultant hydronephrosis. CTU with split contrast bolus.	Asymptomatic: follow-up Symptomatic: Laparoscopic transperitoneal or retroperitoneal repair.
Ovarian vein syndrome	Exclusively seen on right side. Uncommon in nulliparous	Dilated aberrant or thrombosed ovarian vein. Extrinsic vascular impressions upon the ureter at pelvic brim. Proximal hydroureter and normal distal ureter.	Transcatheter embolization. Surgical ligation of the ovarian vein.
Right-sided varicocele due to retroperitoneal mass	Isolated right-sided varicocele extremely rare	Most common etiology is renal cell cancer, followed by RP sarcomas, lymphoma, lymph node enlargement etc.	Surgery with complete resection provides best benefit.

SYNDROM HORNÍ MESENTERICKÉ TEPNY (SMAS)

Wilkie syndrom incidence 0,01–0,3%

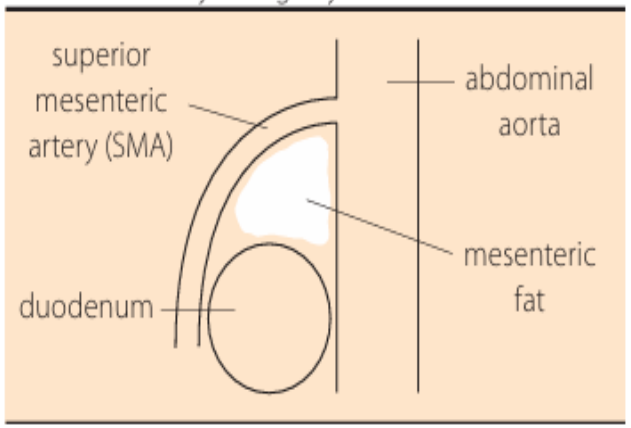
Superior mesenteric artery (SMA) syndrome, also called Wilkie's syndrome or cast syndrome, is a rare disorder in which acute angulation of the SMA causes **compression of the third part of the duodenum** between the SMA and the aorta, leading to **obstruction**. SMA syndrome is an atypical cause of proximal intestinal obstruction, most frequently occurring in young patients who have had an important weight loss.

Table 1 Incidence of superior mesenteric artery syndrome in several populations

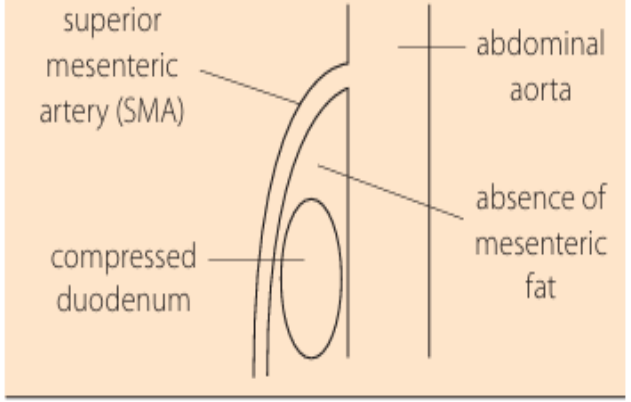
Population	Incidence (%)	Ref.
Acute general hospitals	0.001-0.0052	[18]
Chronic-care hospital	0.097	[18]
Hospital admissions	0.05-2.67	[12,27,28]
Upper gastrointestinal endoscopy	0.48	[29]
Post-scoliosis surgery + cast	0.5-3.4	[19,30-34]
Spinal cord injury	0.53	[21]
Burn injury	1.0-1.12	[22-24]
Anorexia nervosa (admitted)	2.73	[25]
Functional dyspepsia	10.8	[26]

WILKIE SYNDROM INCIDENCE 0,01–0,3%/3:2/10-40 LET

Obrázek 1a. Fyziologický stav



Obrázek 1b. Komprese duodena při sy. Wilkie

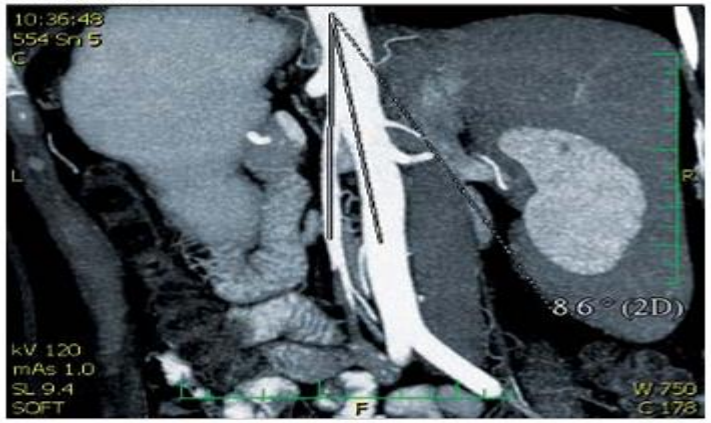


Predispozice

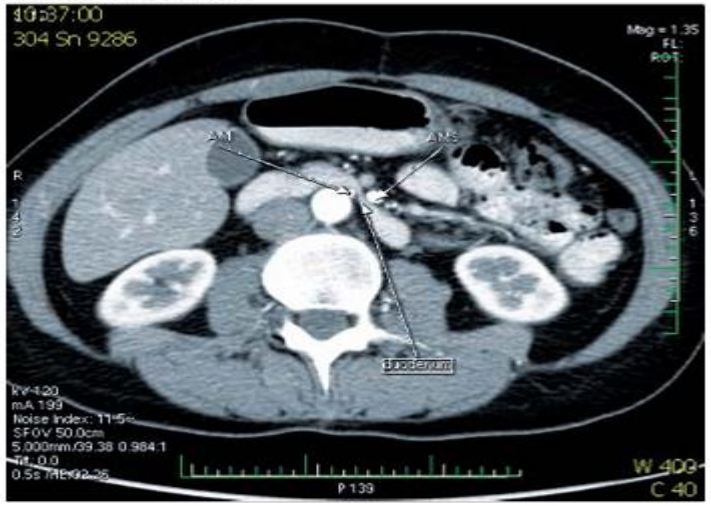
1. **Rychlý váhový úbytek**

2. **Chirurgické výkony (spondylochirurgie)**

Obrázek 2. CT nález pacientky – úhel odstupu SMA z AA



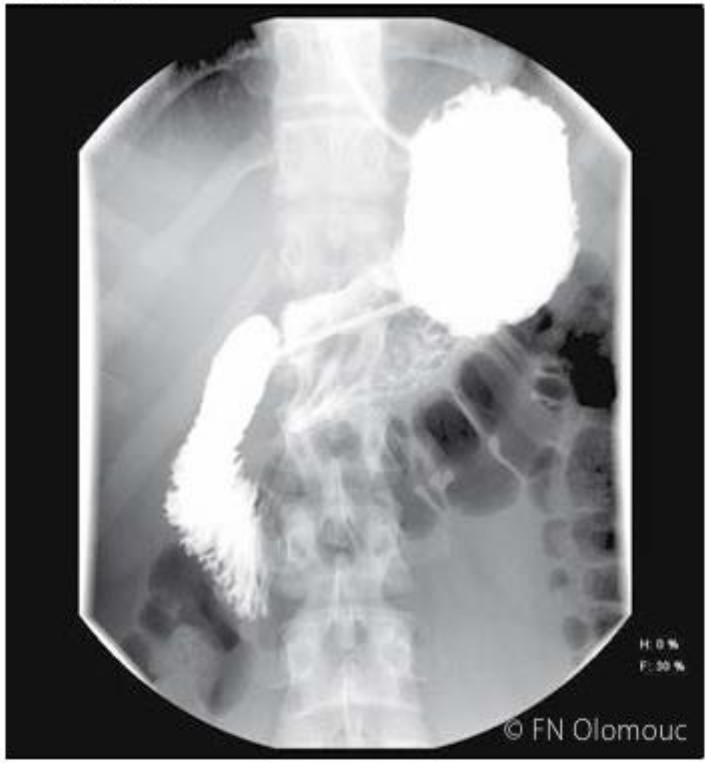
Obrázek 3. CT nález pacientky – komprimované duodenum

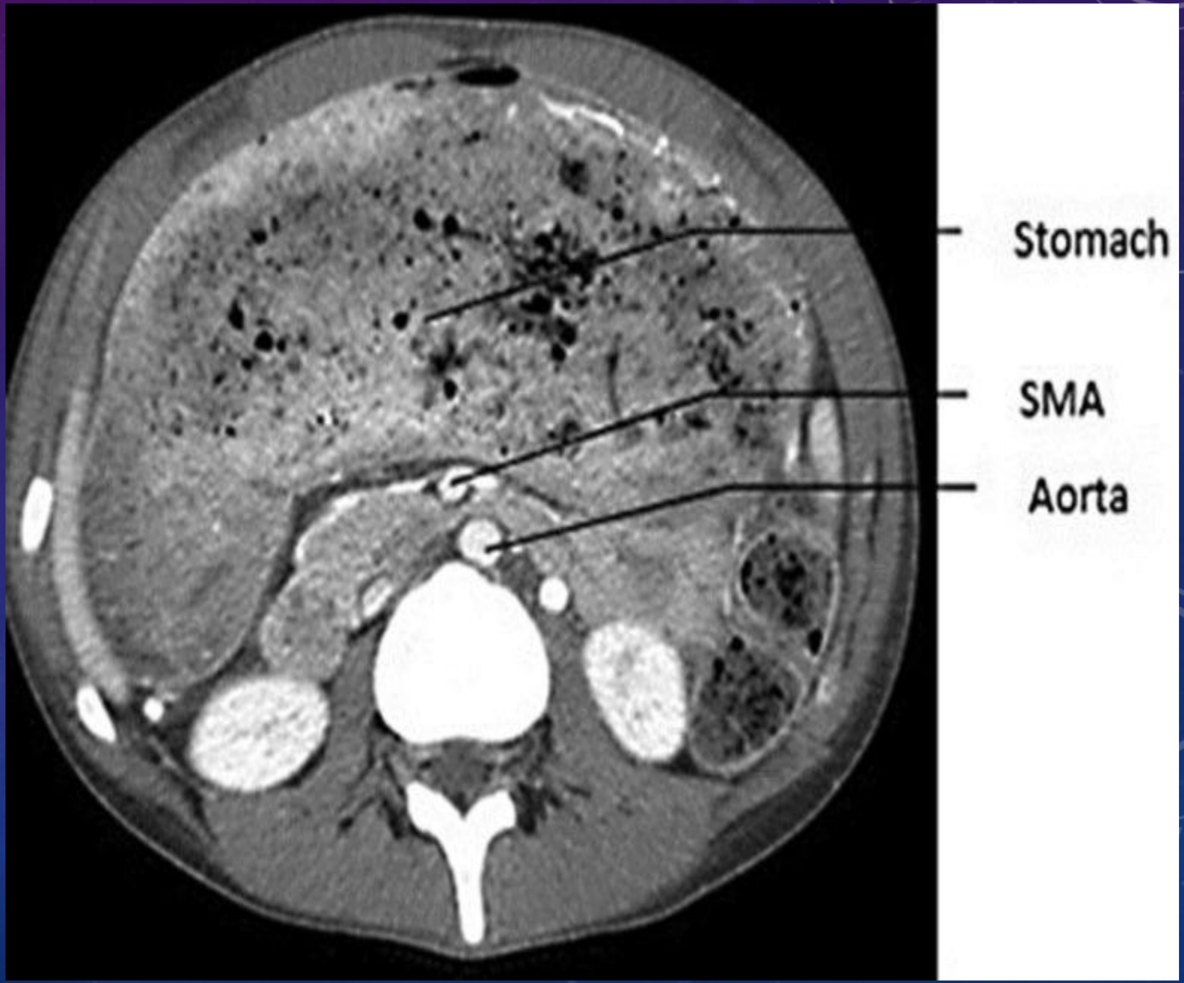


Interní Med. 2011; 13(9): 353–354

CASE REP GASTROENTEROL. 2015 MAY-AUG; 9(2): 194–199.

Obrázek 6. Rtg vyšetření žaludku a duodena, duodenum je dilatované, v oblasti D3 je patrné ukončení kontrastní náplně, další část duodena a jejunum se nezobrazilo





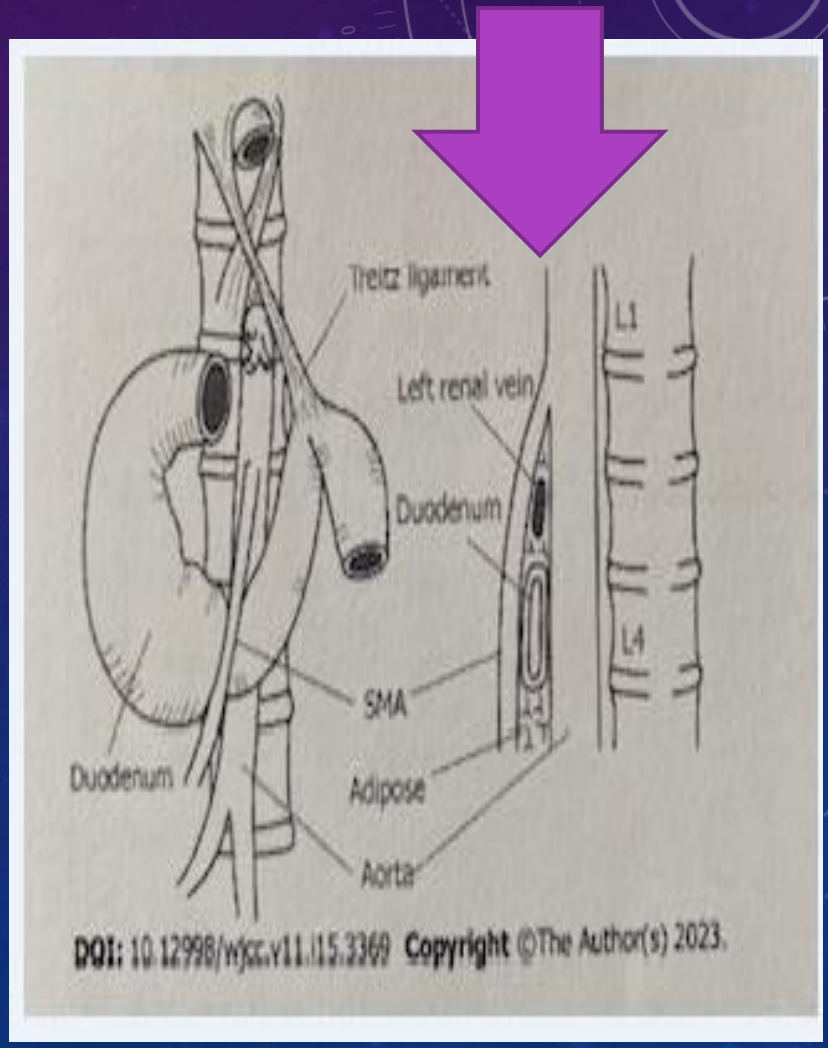
DIAGNOSTIKA SMAS JE ZALOŽENA:

a) zobrazovací metoda (CT CT-AG, MR, UZ)

b) na klinické symptomatologii bolestí a zvracení …aspirace….těžká malnutrice

Úlevová poloha: levo-laterální pokrčené DK k hrudníku

c) potvrzením poruchy pasáže GIT při vyšetření polykacím aktem.



The aorta-SMA angle ranges from **38 to 65**° (25-60) , due to the erect posture, while in quadrupeds, it is nearly a right angle

The main anatomic feature of SMA syndrome is a narrowing of the **aorta-SMA angle to <25° (22),** and as a result, the aortomesenteric **distance decreases to <10 (8) mm,** from normally 10 to 28 (20) mm

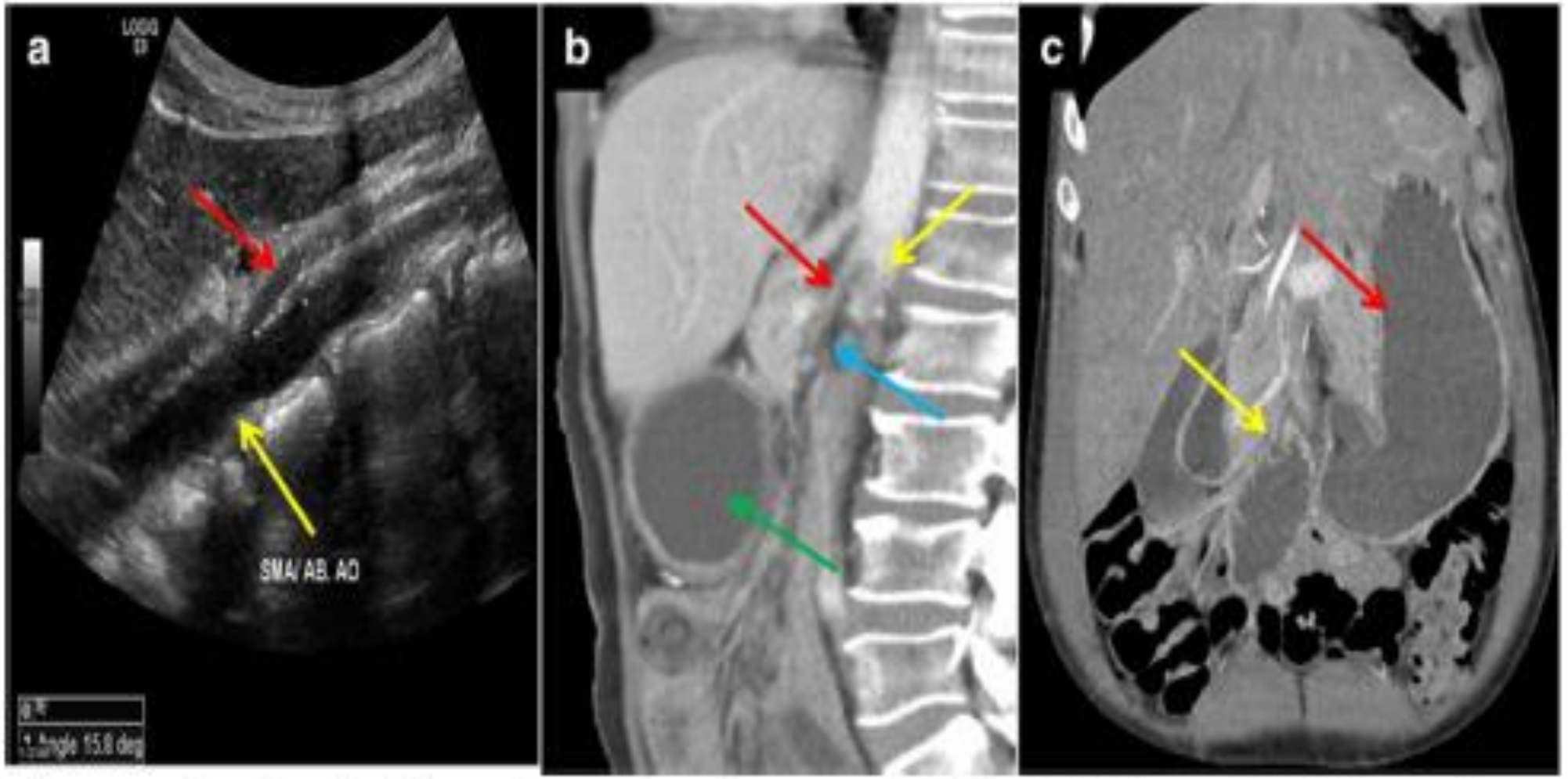


Fig. 3 Superior mesenteric syndrome in different patients. **a** Grayscale ultrasound in mid-sagittal plane shows aortomesenteric angle of 15.8 degrees (measured with calipers, SMA - red arrow, and aorta - yellow arrow). **b** Sagittal CT image showing compression of the 3rd part of the duodenum (blue arrow) between the SMA (red arrow) and aorta (yellow arrow) and distended 2nd part of the duodenum and stomach (green arrow). **c** Coronal CT image of the abdomen showing dilated proximal stomach (red arrow) with abrupt transition at the third part of the duodenum (yellow arrow)

Table 4 Diagnostic modalities for superior mesenteric artery syndrome

Modalities	
Plain film X-ray	
Barium X-ray	
Angiogram	
CT	
	Plain
	Enhanced (3D-CT)
Abdominal ultrasound	
	B-mode
	Doppler-mode
MRI	
	MR angiography
	MR enterography
Endoscopy	
	White light imaging
	Ultrasonography (EUS)
Gastric-emptying scintigraphy	
Multi-channel manometry	

CT: Computed tomography; 3D: Three-dimensional; MRI: Magnetic resonance imaging; EUS: Endoscopic ultrasonography.

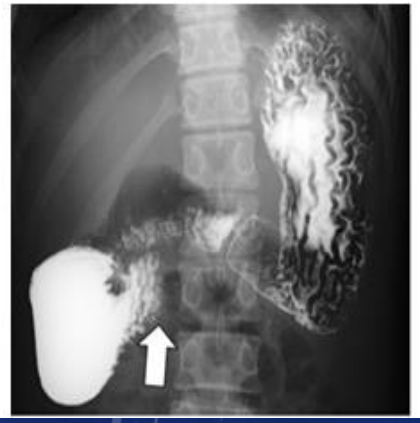


Table 5 Differential diagnoses of superior mesenteric artery syndrome

Disorders mimicking SMA syndrome
Similar symptoms by...
Eating disorder
Anorexia nervosa, anorexia bulimia
CIPO
Peptic ulcer disease
Reflux esophagitis
Functional dyspepsia
Cyclic vomiting syndrome
Pancreatitis
Gastric outlet obstruction
Involvement of duodenum by... (other disorders)
Tubercular infection
Megaduodenum (localized CIPO)
Henoch-Schönlein purpura
Crohn's disease
Celiac disease
Ectopic pancreas
Duodenal diverticula
Duodenal edema
Tumor
Primary or metastatic duodenal cancer, pancreatic cancer, lymphoma, *etc.*
Anatomical abnormality (web, diaphragm)
Foreign body (bezoar, *etc.*)
Extrinsic compression by... (non-SMA)
Aortic artery aneurysm (Aortoduodenal syndrome)
Stent or filter
Mesenteric artery, aorta, IVC, *etc.*
Horseshoe kidney
Lymph node
Abscess
Traumatic false aneurysm

Table 2 Etiology of superior mesenteric artery syndrome

Etiology		Ref.
Congenital		
	Short or high insertion of Treitz ligament	[60]
	Low origin of the SMA	[35]
	Spinal deformity (Scoliosis, Marfan, *etc.*)	[36,37]
	Familial	[38,39]
	Malrotation of SMA and SMV	[40,41]
	Malrotation of intestine	[2]
Body weight loss		
	Diet and obesity surgery (sleeve surgery)	[5,42]
	Eating disorders (anorexia nervosa, anorexia bulimia)	[25,36]
	Malabsorption	[17]
	Malignancy	[17,18,35]
	Tuberculosis	[44]
	Chemotherapy	[45,46]
	Trauma (Burn injury, brain injury, spinal cord injuries, *etc.*)	[22,47,48]
	Neural disorders (ALS, MELAS, paraplegia, cerebral palsy, *etc.*)	[49,50]
	Drug or alcohol abuse	[36]
	Rheumatoid arthritis	[51]
Scoliosis surgery		[19,30-34]
Intestinal surgery (IPAA, colectomy, *etc.*)		[36,52,53]
Aging (bed rest, frail, vascular calcification, *etc.*)		[54,55]
Body cast		[4-6]
"Pseudo-" SMA syndrome		
	Aortic artery aneurysm (Aortoduodenal syndrome)	[56-58]
	Surgery near or around the SMA and 3rd duodenum	[59]

Therapeutic methods	
Conservative therapy	
	Decompression of dilated stomach and duodenum by
	Postural change (left lateral, sitting position)
	Nasal gastric tube suction
	Medication (metoclopramide)
	Gaining weight by
	Giving multiple small feeds
	Feeding tube (nasal gastric or jejunal)
	Total parenteral nutrition
Surgical therapy	
	Anterior transposition of the third part of duodenum
	Gastroduodenostomy
	Gastrojejunostomy
	Duodenojejunostomy
	Strong's procedure (a division of the ligament of Treitz)
	Duodenal lowering
	Ladd's procedure
Endoscopic therapy	
	Lumen-apposing metal stent[1] by
	EUS-guided gastrojejunostomy

[1]Lumen-apposing metal stent: Potential option based on case reports.
EUS: Endoscopic ultrasonography.

CIR ESP. 2020;98(1):48–57

CIRUGÍA ESPAÑOLA

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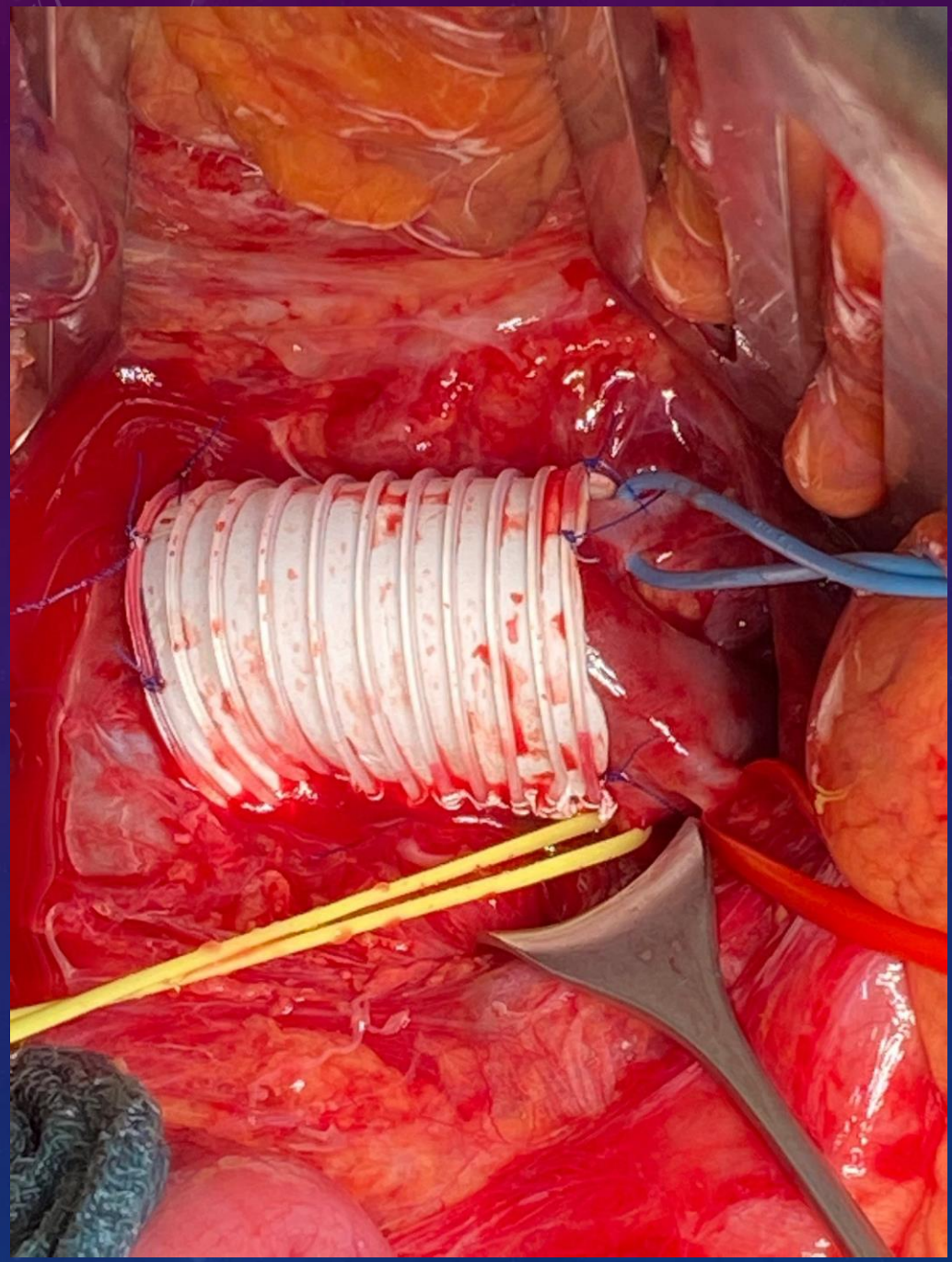
Scientific Letters

Superior Mesenteric Artery Syndrome (Wilkie Syndrome): Analysis of a Series of 5 Cases☆

Síndrome de la pinza aorto-mesentérica (Sind. de Wilkie). Análisis de una serie de 5 casos

Laparoscopic duodenojejunostomy is an effective, minimally invasive treatment, with an acceptable rate of postoperative complications and favorable long-term results, which is why it is considered the treatment of choice

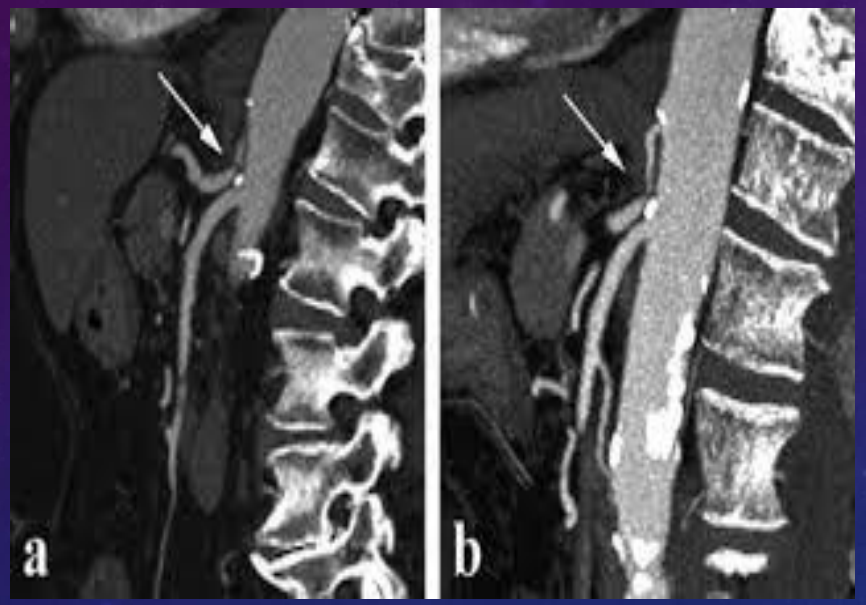
PTFE shield



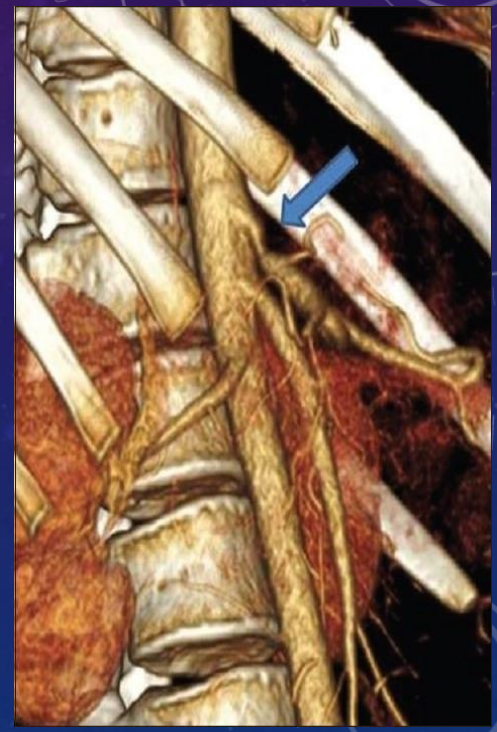
DUNBAR SYNDROM (2/100 000/ 4:1 / VĚK 30-50 LET)

MEDIAN ARCUATE LIGAMENT SYNDROME (OBLAST L1)

CELIAC ARTERY COMPRESSION SYNDROME



- Characteristic Triad:
  - Postprandial Abdominal Pain
  - Weight Loss
  - Epigastric Bruit
- Other Sx:
  - Nausea & Vomiting
  - Diarrhea



Median Arcuate Ligament Syndrome (MALS) - The Operative Review Of Surgery

Hongsakul K, Rookkapan S, Sungsiri J, Tubtawee T. A severe case of median arcuate ligament syndrome with successful angioplasty and stenting. Case Rep Vasc Med. 2012;2012:129870. (License: CC BY-3.0)

Divarci E, Celtik U, Dokumcu Z, Celik A, Ergun O. Laparoscopic Treatment of Median Arcuate Ligament Syndrome: A Rare Cause of Chronic Severe Abdominal Pain. J Indian Assoc Pediatr Surg. 2017 Jan-Mar;22(1):48-50. (License: CC BY-NC-SA-3.0)

85% asymp

- - ischemie
- - iritace pl. coeliacus

- Dx: Duplex US, CTA or MRA
  - Inspiratory & Expiratory Arteriography is Preferred but Often Unable
  - Diagnosis of Exclusion

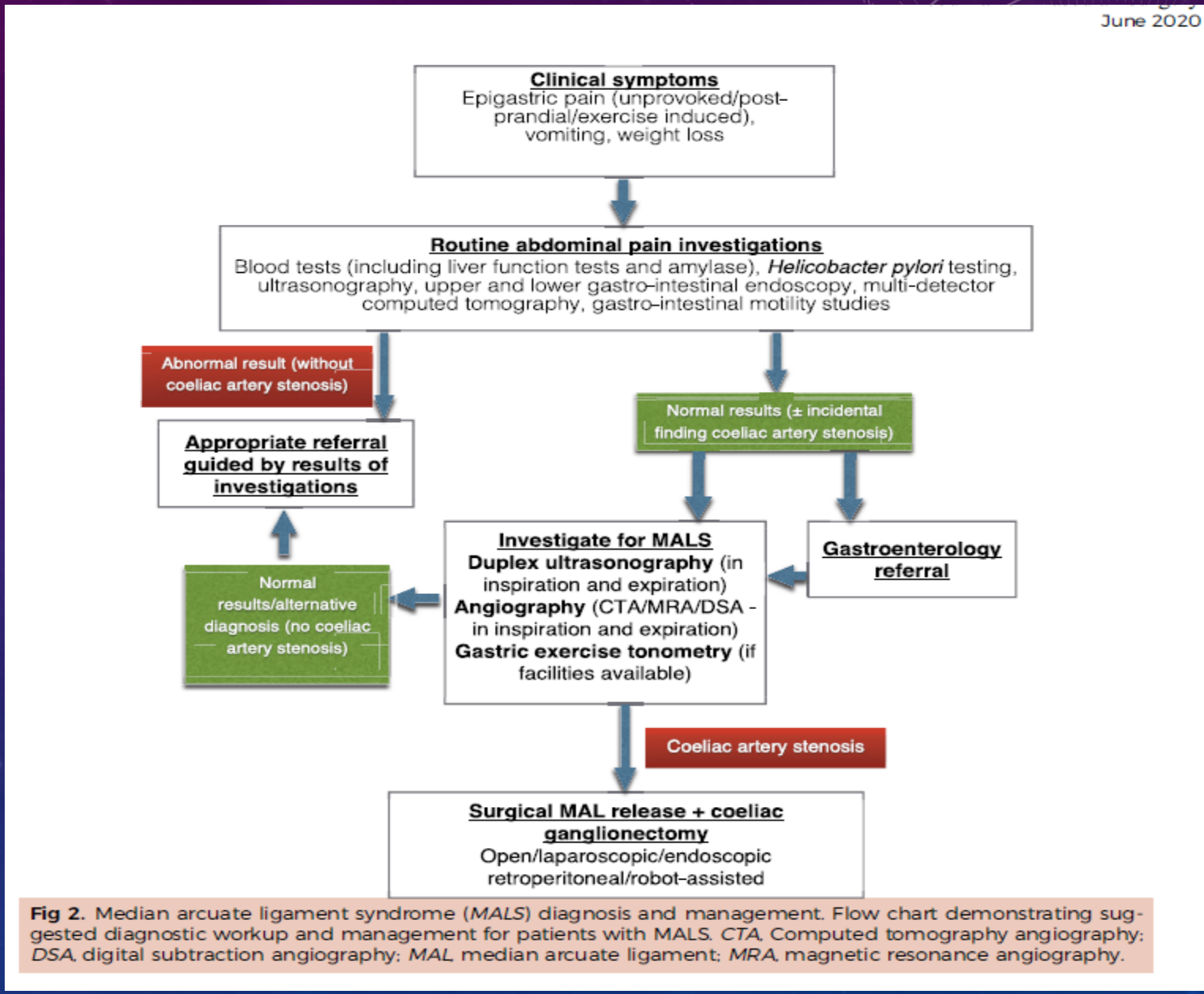


Fig 2. Median arcuate ligament syndrome (*MALS*) diagnosis and management. Flow chart demonstrating suggested diagnostic workup and management for patients with MALS. *CTA*, Computed tomography angiography; *DSA*, digital subtraction angiography; *MAL*, median arcuate ligament; *MRA*, magnetic resonance angiography.

Journal of Vascular Surgery
June 2020

LÉČBA

- Therapeutic intervention of MALS primarily consists of open/laparoscopic surgical **release of the median arcuate ligament,** which essentially results in celiac ganglionectomy.

- In a systematic review of 504 cases, patients undergoing laparoscopic treatment were found to have shorter hospital time and a shorter operative time than open surgical release

- CAVE: aterosklerotické postižení - PTA-stent

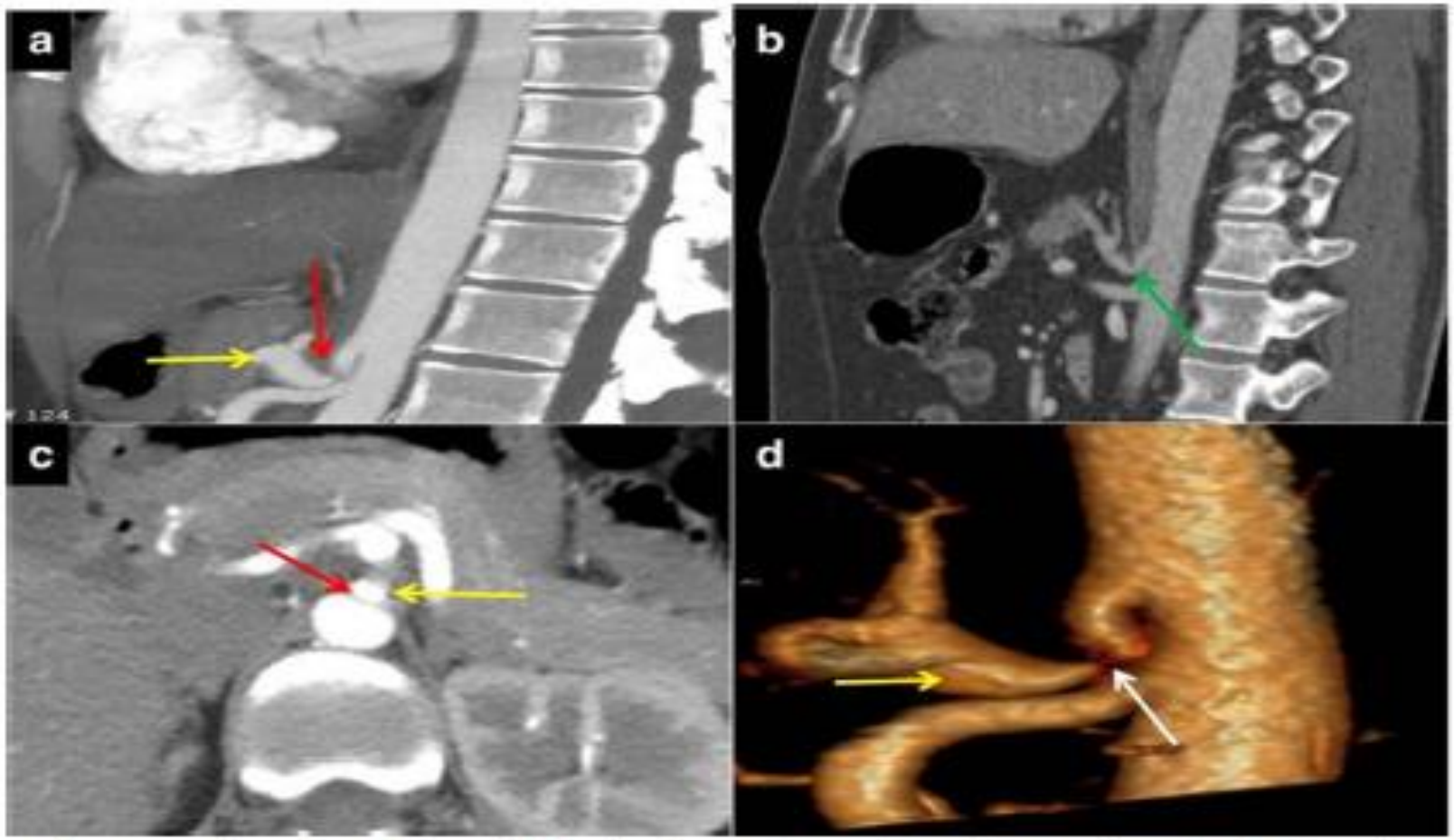


Fig. 1 Median arcuate ligament syndrome in a 47-year-old female with chronic post-prandial upper abdominal pain. CT angiography images of the same patient in **a** sagittal reformatted MIP image of arterial angiogram, **b** sagittal reformatted MPR image of end-inspiratory phase, **c** axial reformatted MIP image of arterial angiogram, and **d** volume rendered 3D reconstructed image. "Hooked" appearance of the proximal celiac artery due to tenting of the superior surface by the median arcuate ligament (red arrows) and post-stenotic dilatation (yellow arrows) of the celiac artery. The characteristic "fish hook" or "J" shape appearance persists on end-inspiratory phase (green arrow on image **b**). The celiac artery stenosis due to extrinsic impression by median arcuate ligament can also be seen with 3D-VR image (white arrow on image **d**)

NUTCRACKER SYNDROME

Heidbreder Journal of Medical Case Reports (2018) 12:214

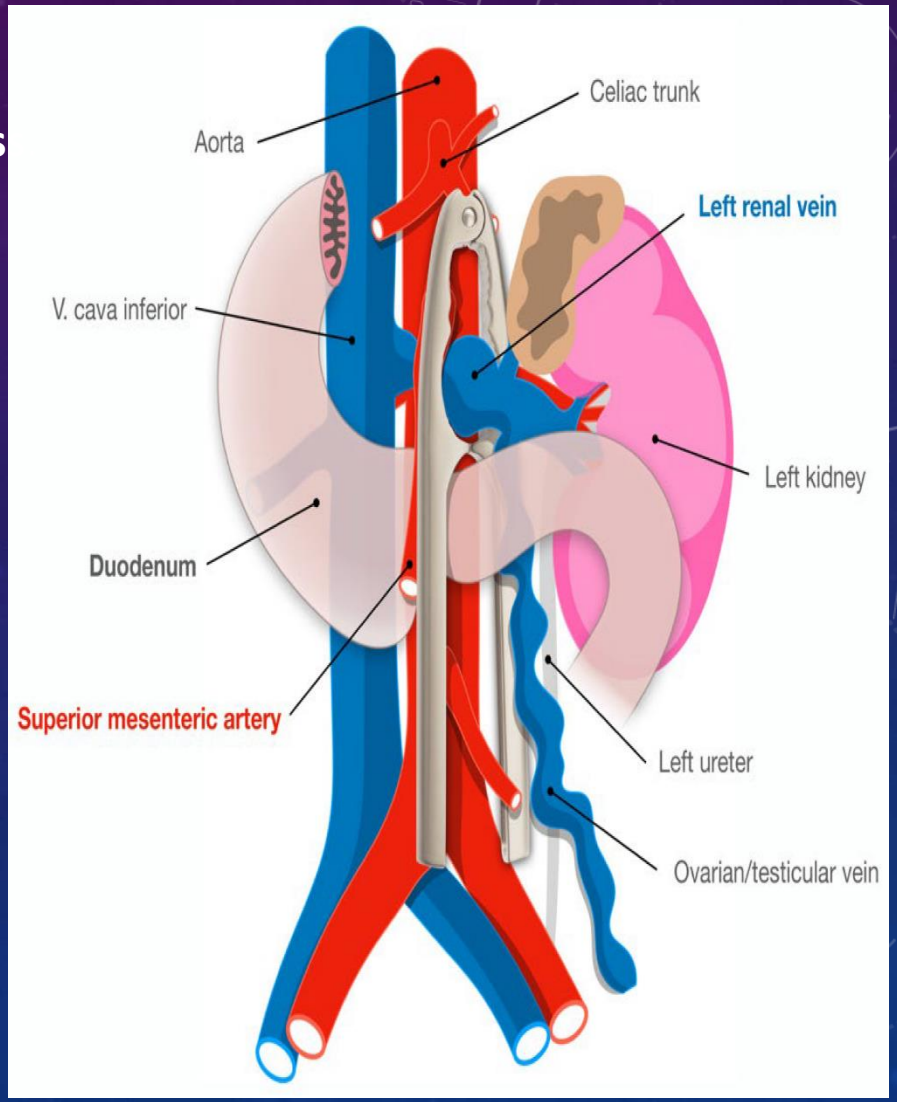
NCS produces **urinary and/or gynecological symptoms** but **generally no weight los**

The manifestations of NCS range from asymptomatic to **left flank pain**, pelvic congestion with left sided varicoceles, hematuria, and proteinuria

A hallmark of NCS is the aggravation of symptoms **with position**, as standing upright causes the viscera to drop down thus pulling the SMA and aggravating the compression

A patient, who first presents with radiographic and clinical evidence of SMAS, could also **simultaneously or sometime thereafter present with clinical symptoms that point to a urologic or gynecologic dysfunction.**

The diagnosis should be suspected if there is evidence of **pelvic congestion, varicocele and/or engorgement of the gonadal vein**, all of which are particularly remarkable findings in younger and nulliparous patients.



Pathologies leading to NCS
Pancreatic neoplasms
Para-aortic lymphadenopathy
Retroperitoneal tumour
Abdominal aortic aneurysm
Overarching testicular artery
Strangulating fibrolymphatic tissue between the aorta and SMA[a, b, c]
High disposition of the LRV[d]
Left renal ptosis resulting in stretching of the left renal vein over the abdominal aorta[e]
Lordosis[e]
Reduced retroperitoneal and mesenteric fat[f, g]
Pregnancy with a gravid uterus compressing the renal vasculature[h]

Less common pathologies leading to NCS based on studies by Menard etal.[a,13], Pastershank etal.[b,28], Neste etal.[c,29], Kurklinsky etal.[d,12], Shokier etal.[e,59], Wendel etal.[f,30], Venkatachalam etal.[g,8], Zhang etal.[h,19], and Itoh etal.[i,31]

Diagnosis

The diagnosis of NCS is confirmed on imaging results, including DUS, CT, magnetic resonance imaging (MRI), phlebography, and intravascular ultrasound (IVUS), (illustrated in Fig. 3).7, 11, 33, 45 However, the diagnosis of NCS can still be challenging to confirm based solely on imaging; the presenting clinical signs and symptoms are therefore paramount when considering management options.12, 46

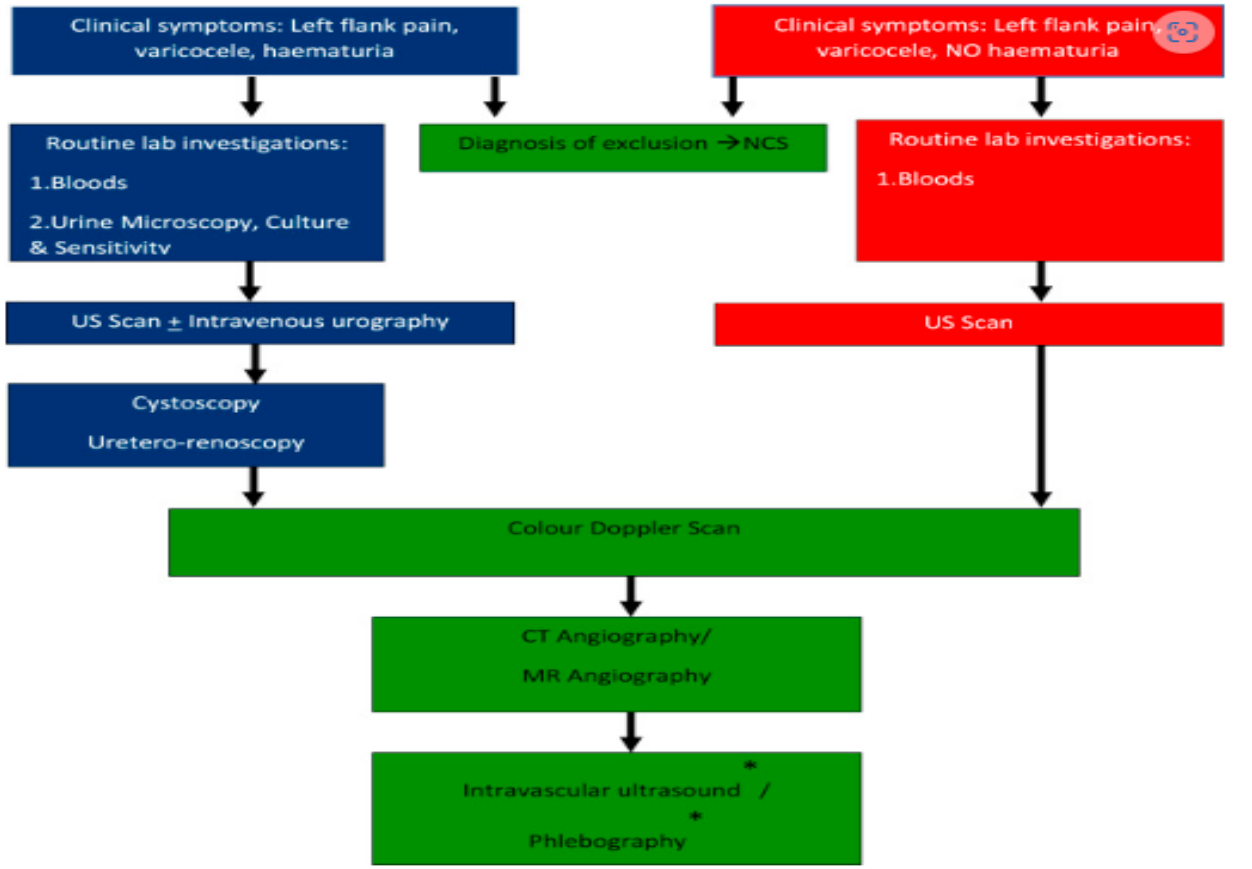


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Figure 3. Flow chart showing how NCS is diagnosed following a pathway from initial symptomatic presentation to a series of tests from the least invasive to most invasive. Adapted from figures by He etal. (2014)[11] and Ahmed etal. (2006).[10] *=Gold standard.

NUTCRACKER SYNDROME

Heidbreder Journal of Medical Case Reports (2018) 12:214

Mayo Clin Proc. • June 2010;85(6):552-559

Cystoscopy may identify **a left ureteral origin**

The diagnosis can be made by Doppler ultrasound, CT scan, or MRI where a **dilated vein with a delayed washout and pelvic varicosities** in the right clinical setting are highly suggestive.

LRV phlebography is an invasive but **conclusive method** for confirming the elevated pressure in the LRV

Insufficient knowledge of the natural history of these conditions results in uncertainty in treatment selection and diagnostic criteria.

The term nutcracker syndrome should be reserved for patients with characteristic clinical symptoms associated with demonstrable nutcracker morphologic features.

NUTCRACKER PHENOMENON AND NUTCRACKER SYNDROME

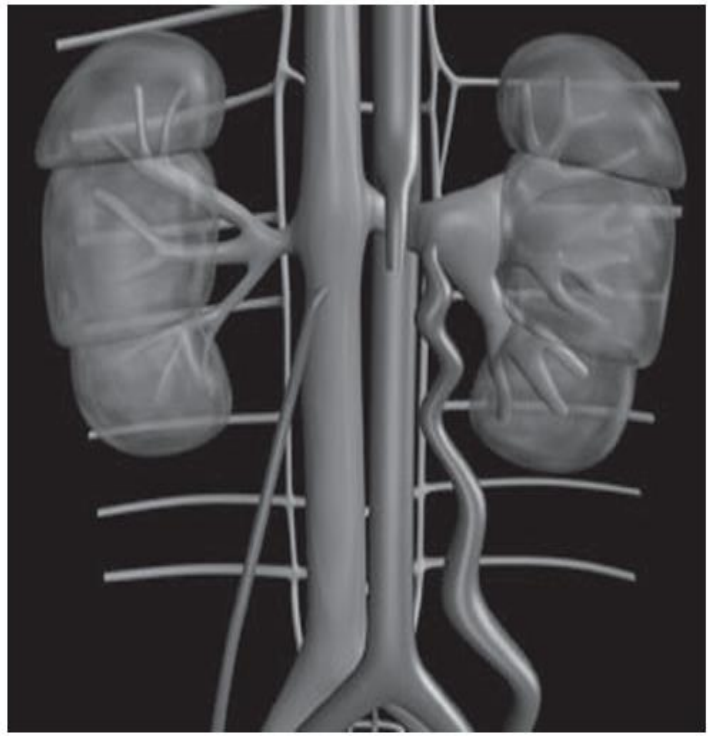


FIGURE 1. Schematic representation of nutcracker phenomenon. Hilar portion of the left renal vein and the gonadal vein are distended. Distended lumbar and azygous collaterals may be seen in some cases.

TREATMENT NCS

- Management options range **from observation to nephrectomy**, depending on the severity of symptoms.
- Conservative treatment is recommended for mild hematuria.
- For patients younger than 18 years, the best option is a conservative approach with observation for at least 2 years because as many as 75% of patients will have complete resolution of hematuria.
- Angiotensin inhibitors may be helpful in improving orthostatic proteinuria in patients with NCS.

TREATMENT
LONG-TERM NCS FOLLOW-UP DATA ARE LACKING.

- Most interventions **aim to decrease LRV hypertension**, but others are directed against pelvic venous reflux.
- A variety of surgical approaches have been used, including medial nephropexy with excision of renal varicosities,

LRV bypass,

LRV transposition with or without Dacron wedge insertion between SMA and aorta

SMA transposition,

renal-to-IVC shunt

renal autotransplant, gonadocaval bypass,

nephrectomy for persistent hematuria.

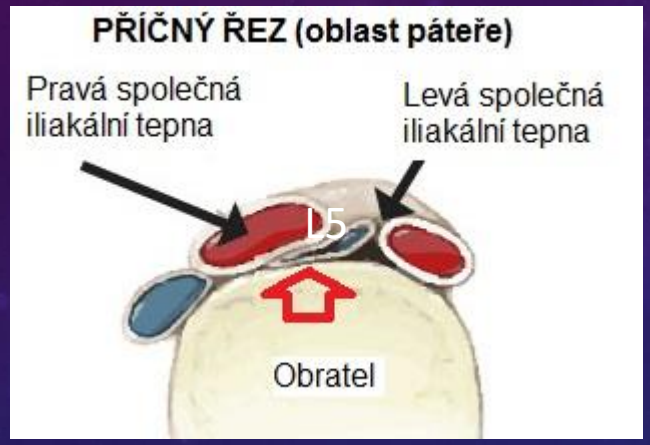
- External stenting with ringed **polytetrafluoroethylene graft (PTFE stent)** interposition around the LRV and intravascular stenting have been applied relatively recently.
- **Endovascular treatment of NCS is becoming a preferred** modality owing to its minimally invasive nature

Treatment NCS

Disadvantage of EVS is that patients must be on anticoagulation and antiplatelet therapy for a short time thereafter.

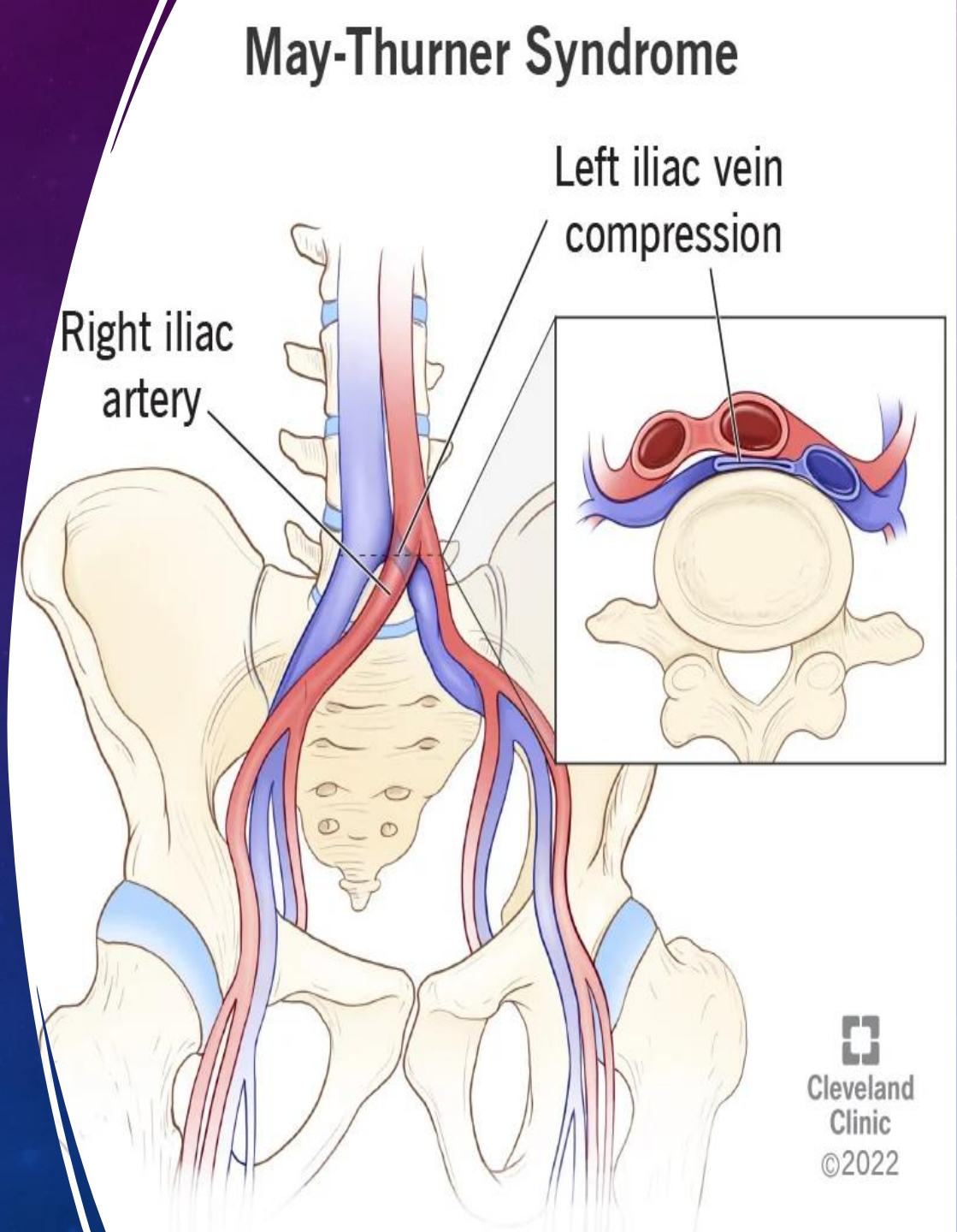
A recommended regimen is low molecular weight heparin for 3 days after EVS,
30 days of clopidogrel and ≥3 months of aspirin

May-Thurnerův syndrom (iliac vein compression syndrome, May-Thurner syndrome)
- způsoben kompresí společné pánevní žíly (vena iliaca communis), která je utlačována společnou pánevní tepnou (arteria iliaca communis).



V praxi MTS + POTs + EDS……..+ wilkie + dunbar….

EDS-related connective tissue abnormalities and the resulting increased risk for vein stenosis may interact synergistically to produce more severe haemodynamic and autonomic POTS symptoms.



LÉČBA MTS

- Prior to the year 2000, 75% of procedures for MTs treat ment involved open surgery and 25% were performed with an endovascular approach.
- In contrast, between 2000-2014 there was a significant shift towards **minimally invasive treatment**, with only 4.1% of patients having open surgery.
- The current treatment of choice for MTs is venography- and iVus-guided, **endovascular stenting.**
- Despite differences in presentation and treatment of thrombotic and non-thrombotic MTs, both patient groups appear to benefit from high stent patency rates and a high degree of symptom relief with iliac vein stenting.
- Left common iliac vein stenting can mitigate POts symptoms by decreasing lower extremity venous pooling and subsequently improve downstream sympathetic nervous system hyperactivation

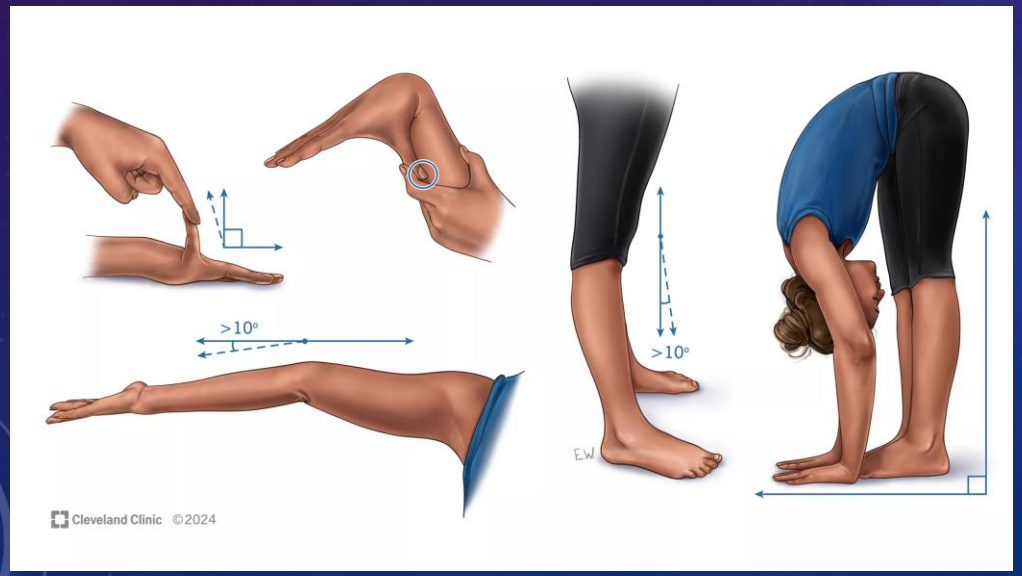
The Journal of Cardiovascular surgery 2021

EHLERS - DUNLOSŮV SYNDROM

- vážné, vzácné dědičné onemocnění pojivové tkáně
- způsobené porušenou tvorbou kolagenu

Příznaky:

- postižení pohybového aparátu – silná bolest kloubů, svalů, často vyžadující opiátovou medikaci
- hypermobilita kloubů, vývojové dysplázie, skolióza, hyperlordóza, hyperkyfóza, organoptóza
- cévy – aneuryzmata, disekce aorty, syndrom vaskulární komprese (louskáčkův syndrom, MayThurner, Dunbar, Wilkie)
- nervový systém – neuropatie, POTS, únava, porucha aktivace žírných buněk (MCAS), zhoršené hojení



EHLER - DANLOS

- Ehlers–Danlos syndrome (EDS) was first recognized in the time of Hippocrates in the fourth century BC.
- EDS is the most common non-inflammatory connective tissue disorder featuring joint hypermobility, with the hypermobile EDS (hEDS) subtype representing 80–90% of the burden of disease
- hEDS is now recognized as part of the “hypermobility spectrum disorders” (HSD), which are characterized by varying articular and extra-articular involvement and impact on quality of life.

The International Consortium on Ehlers-Danlos Syndromes & Related Disorders
In Association with The Ehlers-Danlos Society

Diagnostic Criteria for Hypermobile Ehlers-Danlos Syndrome (hEDS)

This diagnostic checklist is for doctors across all disciplines to be able to diagnose EDS

Distributed by The Ehlers Danlos Society.

Patient name: _____ DOB: _____ DOV: _____ Evaluator: _____

The clinical diagnosis of hypermobile EDS needs the simultaneous presence of all criteria, 1 ***and*** 2 ***and*** 3.

CRITERION 1 – Generalized Joint Hypermobility

One of the following selected:

- ☐ ≥6 pre-pubertal children and adolescents
- ☐ ≥5 pubertal men* and women* to age 50
- ☐ ≥4 men* and women* over the age of 50

Beighton Score: _____/9

If Beighton Score is one point below age- and sex-specific cut off, two or more of the following must also be selected to meet criterion:

- ☐ **Can you now (or could you ever) place your hands flat on the floor without bending your knees?**
- ☐ Can you now (or could you ever) bend your thumb to touch your forearm?
- ☐ As a child, did you amuse your friends by contorting your body into strange shapes or could you do the splits?
- ☐ As a child or teenager, did your shoulder or kneecap dislocate on more than one occasion?
- ☐ Do you consider yourself "double jointed"?

CRITERION 2 – Two or more of the following features (A, B, or C) must be present

Feature A (five must be present)

- ☐ **Unusually soft or velvety skin**
- ☐ Mild skin hyperextensibility
- ☐ Unexplained striae distensae or rubae at the back, groins, thighs, breasts and/or abdomen in adolescents, men or pre-pubertal women **without a history of significant gain or loss of body fat or weight**
- ☐ Bilateral piezogenic papules of the heel
- ☐ Recurrent or multiple abdominal hernia(s)
- ☐ Atrophic scarring involving at least two sites and without the formation of truly papyraceous and/or hemosideric scars as seen in classical EDS
- ☐ **Pelvic floor, rectal, and/or uterine prolapse in children, men or nulliparous women without a history of morbid obesity or other known** predisposing medical condition
- ☐ Dental crowding and high or narrow palate
- ☐ **Arachnodactyly, as defined in one or more of the following:**
  (i) positive wrist sign (Walker sign) on both sides, (ii) positive thumb sign (Steinberg sign) on both sides
- ☐ Arm span-to-height ratio ≥1.05
- ☐ Mitral valve prolapse (MVP) mild or greater based on strict echocardiographic criteria
- ☐ Aortic root dilatation with Z-score >+2

Feature A total: _____/12

Feature B

- ☐ **Positive family history; one or more first-degree relatives independently meeting the current criteria for hEDS**

Feature C (must have at least one)

- ☐ Musculoskeletal pain in two or more limbs, recurring daily for at least 3 months
- ☐ Chronic, widespread pain for ≥3 months
- ☐ Recurrent joint dislocations or frank joint instability, in the absence of trauma

CRITERION 3 – All of the following prerequisites MUST be met

1. Absence of unusual skin fragility, which should prompt consideration of other types of EDS
2. Exclusion of other heritable and acquired connective tissue disorders, including autoimmune rheumatologic conditions. In patients with an acquired CTD (*e.g.* Lupus, Rheumatoid Arthritis, etc.), additional diagnosis of hEDS requires meeting both Features A and B of Criterion 2. Feature C of Criterion 2 (chronic pain and/or instability) cannot be counted toward a diagnosis of hEDS in this situation.
3. Exclusion of alternative diagnoses that may also include joint hypermobility by means of hypotonia and/or connective tissue laxity. Alternative diagnoses and diagnostic categories include, but are not limited to, neuromuscular disorders (*e.g.* Bethlem myopathy), other hereditary disorders of the connective tissue (e.g. other types of EDS, Loeys-Dietz syndrome, Marfan syndrome), and skeletal dysplasias (*e.g.* osteogenesis imperfecta). Exclusion of these considerations may be based upon history, physical examination, and/or molecular genetic testing, as indicated.

Diagnosis: _____

v9

*Sex assigned at birth. Currently, we do not have data to provide specific additional guidance to individuals on changes in joint hypermobility following treatments that alter sex hormone levels.

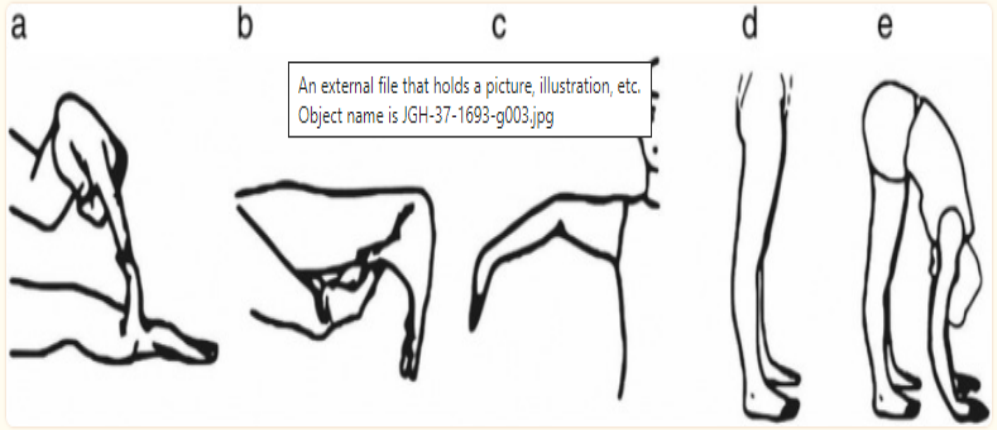


Figure 1

Beighton scoring system measures joint hypermobility on a 9-point scale. Joints assessed (left to right) include (a) passive dorsiflexion of fifth finger ≥ 90° (one point per side); (b) passive apposition of the thumb to ipsilateral forearm (one point per side); (c) hyperextension of the elbow ≥ 10° (one point per side); (d) hyperextension of the knee ≥ 10° (one point per side); and (e) spinal assessment (one point if both palms reach the floor when bending over with knees locked in extension and feet together). Redrawn from Malfait *et al.* (2017) with permission.

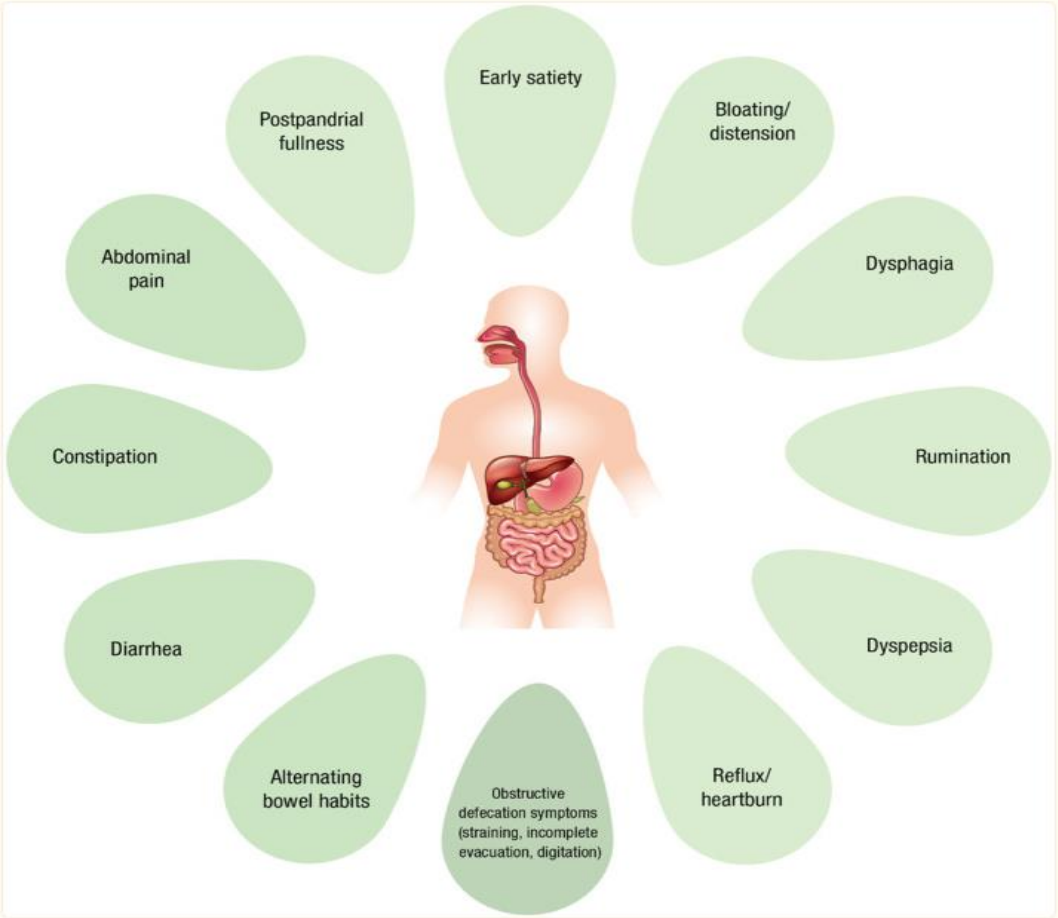


Figure 3

Various gastrointestinal symptoms have been reported to occur significantly more often in patients with HSD/hEDS compared with non-HSD/hEDS controls. See Tables 2 and 3.

Hypermobilní ED - HEDs

POTS v 80%, únava, gastrointestinální obtíže, modřiny, inkontinence, instabilita kloubů, prolapsy chlopní, orgánů, organoptóza……. Raynaudův fenomén,

Vaskulární syndromy + GIT obtíže

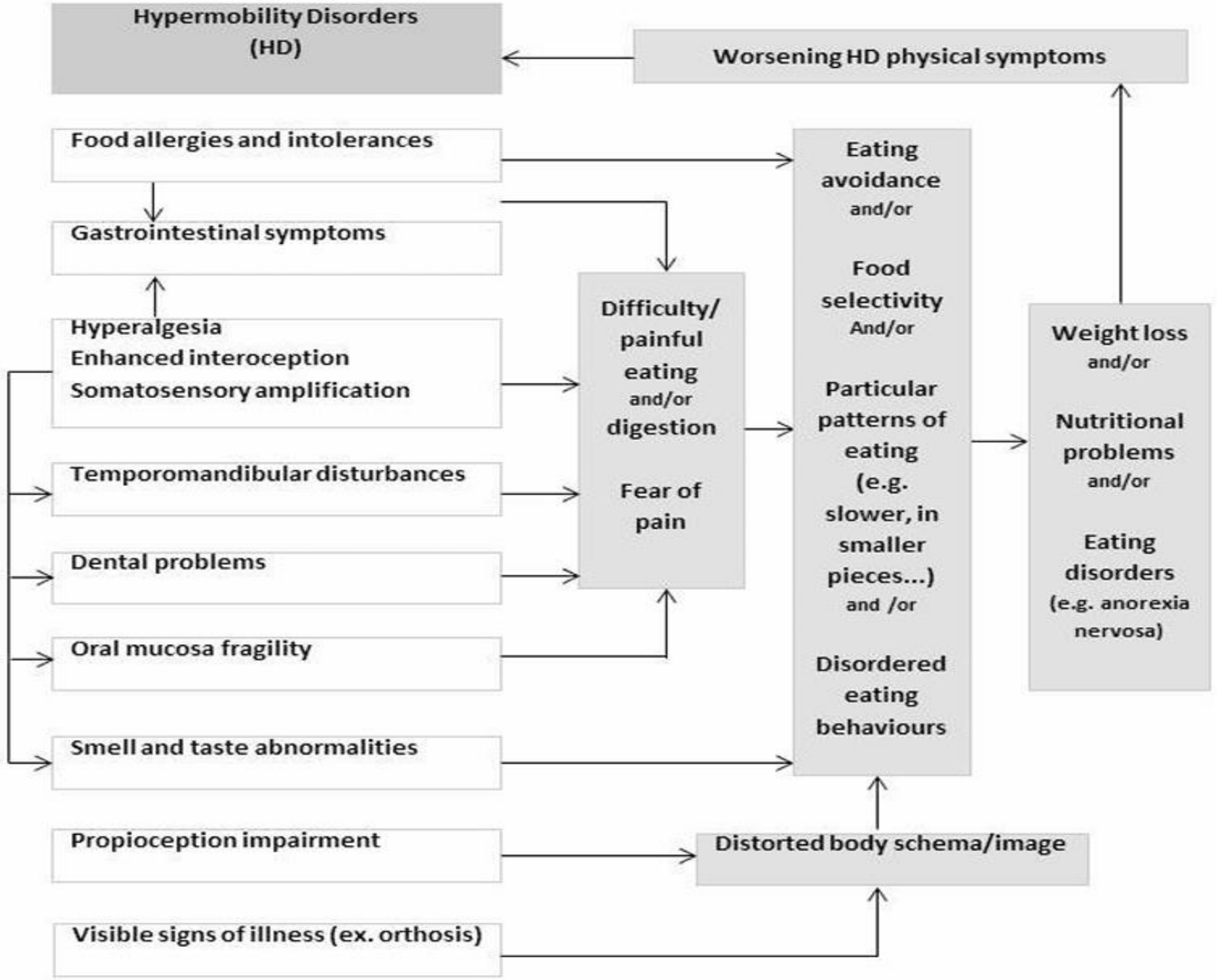


Figure 1 Potential contributors to developing and maintaining disturbed eating behaviours and significant weight loss seen in HD, adapted fro Bulbena *et al*.[41]

POTS – POSTURÁLNÍ ORTOSTATICKÁ TACHYKARDIE

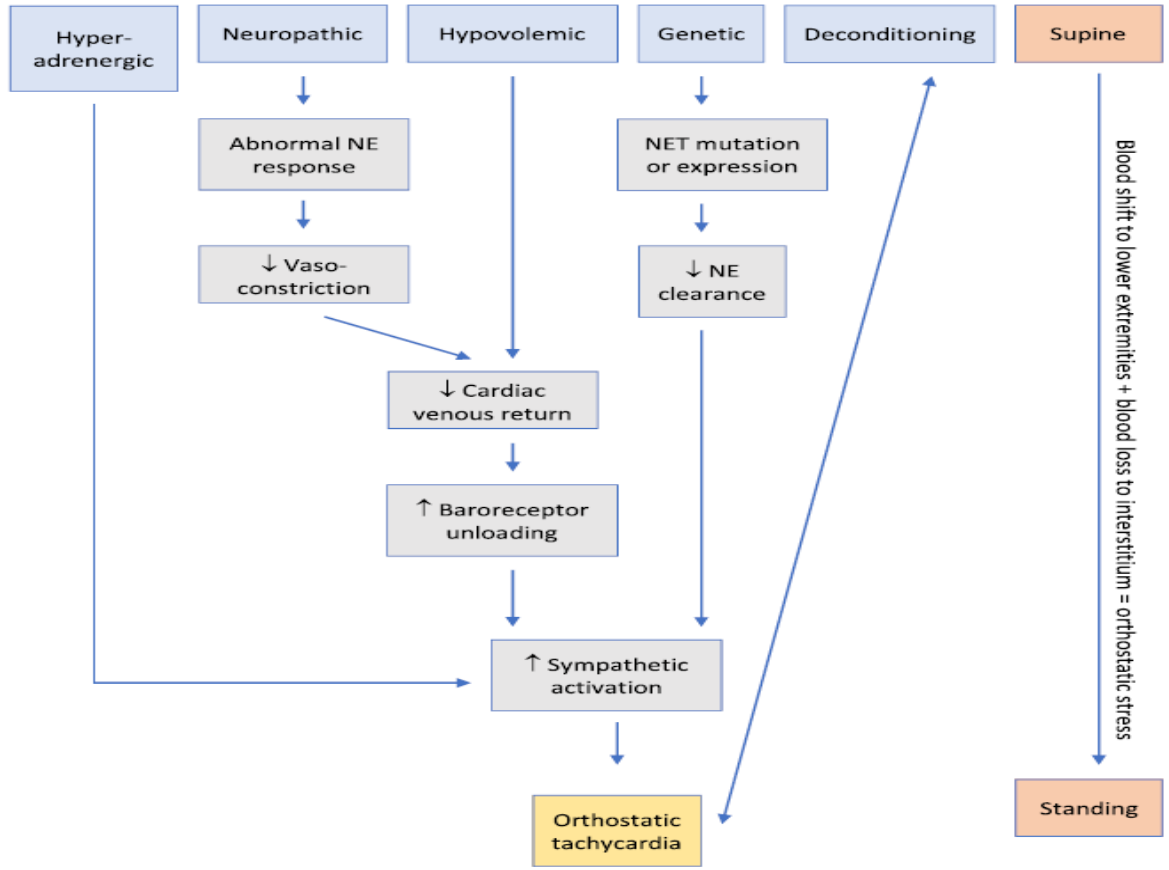


Fig. 1 Convergence of common POTS pathophysiological theories, partially adapted from Arnold et. al. [38]. This figure focuses solely on the principal symptoms of POTS, orthostatic tachycardia, omitting other symptoms of POTS. (*NE* norepinephrine, *NET* norepinephrine transporter)

Table 6 2015 Diagnostic criteria for postural orthostatic tachycardia syndrome. Adapted from Sheldon et al. [44]

Criteria: All 3 must be met and no other cause of tachycardia present
1. Heart rate increase: ≥30 bpm in adults ≥40 bpm in adolescents age 12–19
2. Absence of orthostatic hypotension
3. Symptoms of chronic orthostatic intolerance ≥6 months

Recommendations for Treatment of Postural Orthostatic Tachycardia Syndrome

	Class	Level
A regular, structured, and progressive exercise program for patients with POTS can be effective.	IIa	B-R
It is reasonable to treat patients with POTS who have short-term clinical decompensations with an acute intravenous infusion of up to 2 L of saline.	IIa	C
Patients with POTS might be best managed with a multidisciplinary approach.	IIb	E
The consumption of up to 2–3 L of water and 10–12 g of NaCl daily by patients with POTS may be considered.	IIb	E
It seems reasonable to treat patients with POTS with fludrocortisone or pyridostigmine.	IIb	C
Treatment of patients with POTS with midodrine or low-dose propranolol may be considered.	IIb	B-R
It seems reasonable to treat patients with POTS who have prominent hyperadrenergic features with clonidine or alpha-methyldopa.	IIb	E
Drugs that block the norepinephrine reuptake transporter can worsen symptoms in patients with POTS and should not be administered.	III	B-R
Regular intravenous infusions of saline in patients with POTS are not recommended in the absence of evidence, and chronic or repeated intravenous cannulation is potentially harmful.	III	E
Radiofrequency sinus node modification, surgical correction of a Chiari malformation type I, and balloon dilation or stenting of the jugular vein are not recommended for routine use in patients with POTS and are potentially harmful.	III	B-NR

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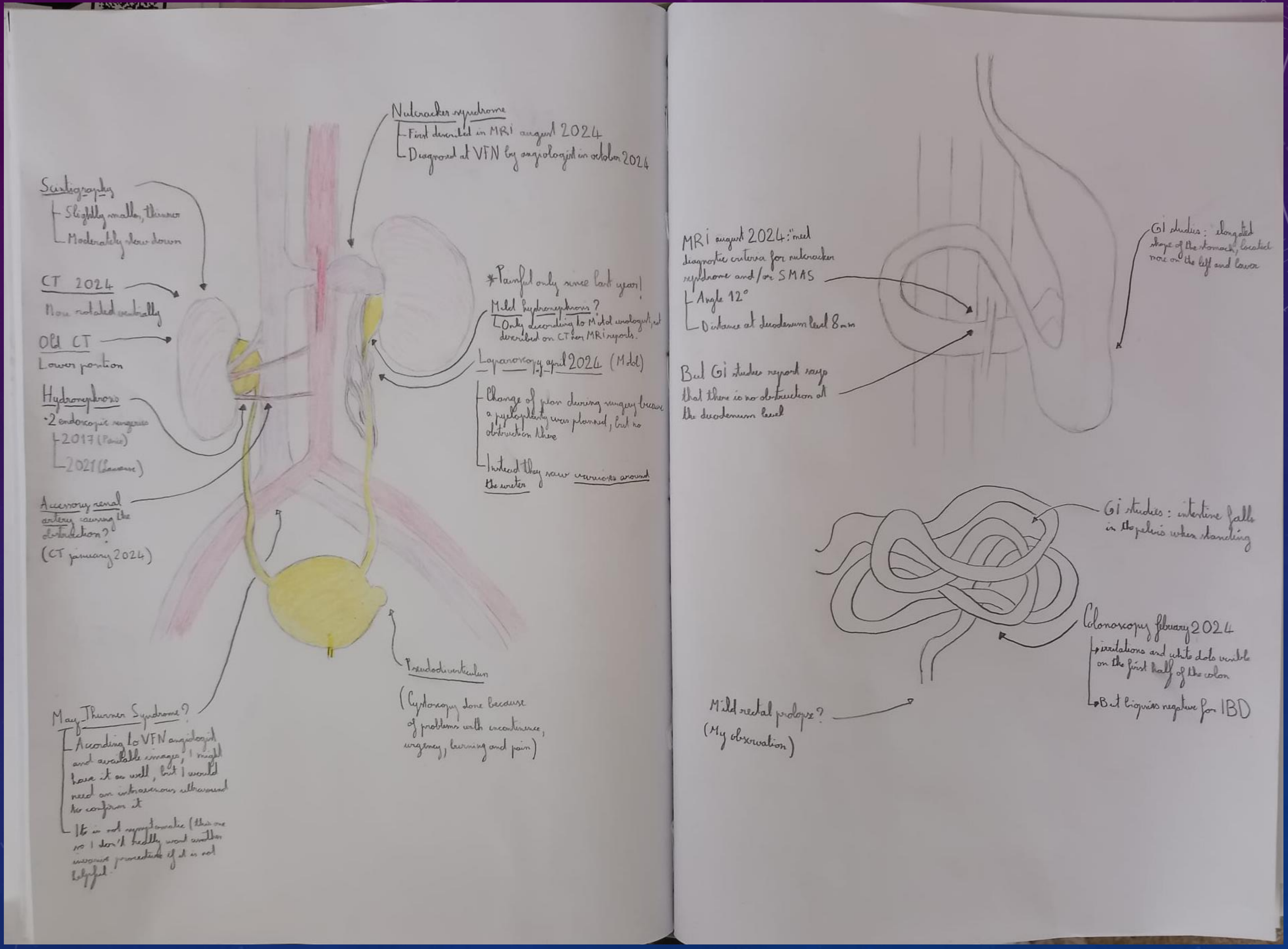
CO LZE JESTĚ NALÉZT…….

1. Mast Cell Activation Syndrome (MCAS)
2. Small Fiber Neuropathy (SFN)
3. Eagle's Syndrome (ES)
4. Slipping Rib Syndrome (SRS)
5. Bertolottis Syndrome
6. Craniocervical Instability (CCI)
7. Postural Orthostatic Tachycardia Syndrome (POTS)
8. Chronic Fatigue Syndrome (CFS)
9. Chiari Malformation (type 1)
10. Irritable Bowel Syndrome (IBS)
11. Tethered Cord Syndrome (TCS)
12. Thoracic Outlet Syndrome (TOS)
13. Tarlov Cyst Syndrome
14. Loin Pain Hematuria Syndrome (LPHS)
15. Pectus Excavatum
16. Anterior Cutaneous Nerve Entrapment Syndrome (ACNES)

various non-musculoskeletal manifestations
neurological (e.g. headaches),
psychiatric and neurodevelopmental (e.g. mood disorders, anxiety, sleep disturbances)
cardiorespiratory (e.g. palpitations, chest pain, and dyspnea),
autonomic (e.g. syncope, postural instability, thermoregulatory difficulties),
urogynecological (e.g. prolapse, urinary incontinence, and dyspareunia),
gastroenterological

SOUBOR IKEM OD 8/2023 – 5/2024 N=15

Wilkiho syndrom	Dunbarův syndrom	Renální syndrom louskáčku	Ehler-danlosův syndrom	POTS	May-thurnerův syndrom	hypermobilita	nutrice
✓	✓	✓	✓	✓	✓	✓	hydratace
✓						✓	DPV
✓			✓ susp	×		✓	neg
✓						✓	neg.
			✓ susp.				neg.
✓			✓	✓		✓	sipping
✓			neg.			neg.	NJS/…..DPV
✓						✓	neg.
neg.			✓	✓	✓	✓	hydratace



Nutcracker syndrome
First described in MRI august 2024
Diagnosed at VFN by angiologist in october 2024
Scintigraphy
Slightly smaller, thinner
Moderately slow down
CT 2024
Now rotated ventrally
Old CT
Lower position
Hydronephrosis
· 2 endoscopic surgeries
2017 (Paris)
2021 (Lausanne)
Accessory renal artery causing the obstruction?
(CT january 2024)
* Painful only since last year!
Mild hydronephrosis?
Only according to Motol urologist, not described on CT nor MRI reports.
Laparoscopy april 2024 (Motol)
Change of plan during surgery because a pyeloplasty was planned, but no obstruction there
Instead they saw varicoses around the ureter
Pseudodiverticulum
(Cystoscopy done because of problems with incontinence, urgency, burning and pain)
May-Thurner Syndrome?
According to VFN angiologist and available images, I might have it as well, but I would need an intravenous ultrasound to confirm it
It is not symptomatic (this one) so I don't really want another invasive procedure if it is not helpful
MRI august 2024: meet diagnostic criteria for nutcracker syndrome and/or SMAS
Angle 12°
Distance at duodenum level 8 mm
But GI studies report says that there is no obstruction at the duodenum level
GI studies: elongated shape of the stomach, located more on the left and lower
GI studies: intestine falls in the pelvis when standing
Colonoscopy february 2024
irritations and white dots visible on the first half of the colon
But biopsies negative for IBD
Mild rectal prolapse?
(My observation)

Jak vypadá pacient aneb 1. návštěva u nás v nutriční ambulanci, typické obtíže:

- mladá slečna s bolestí břicha, únavou, vyčerpáním
- snaží se jíst, ale nejde to, má bolesti břicha, nauzeu, někdy zvrací
- hubne, celkově se necítí dobře, neprospívá
- až obsedantně – kompulzivní chování, selfmonitorace, zjišťování medicínských termínů, vyžadování dalších vyšetření, intervencí
- psychická deprivace
- mnohdy vs. psychiatrická diagnóza, porucha příjmu potravy, anorexie?
- chodí k lékaři, ten neví, co s ní, často jí obtíže nevěří
- začarovaný kruh, nikam to extra nevede, potíže jsou, ale zdají se být neřešitelné…

ZÁVĚR

- Kompresní syndromy (CS) jsou sice vzácnější problematikou, ale klinicky složitou…..
- Kombinace možných syndromů + dalších symptomů – vyloučení hypermobilního EHS (S ANEBO BEZ KOMPRESNÍCH SYNDROMŮ)
- PRO PROGRAM DPV je důležité , že zejména Wilkieho syndrom a POTs je možné léčit DPV (po předchozím pokusu o zlepšení enterální příjmu……
- Systematická péče a program v ČR není ani pro EDS ani pro kompresní syndromy
- Psychologická podpora - léčba – dispenzarizace –
- REHABILITACE