

When Chest Pain Hides a Tumor: the Power of Red Cardiovascular Flags to Unmask a Cardiac Hemangioma in an Adolescent Athlete

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Case report. A 16-year-old male adolescent athlete consulted his primary pediatrician for retrosternal pain occurring immediately after intense physical activity. The pain was described as a low-intensity tightness, accompanied by dizziness, lasting a few minutes, without any other cardiovascular symptoms. The adolescent was asymptomatic at rest and otherwise healthy, with a negative cardiovascular family history.

The clinical examination revealed a new 3/6 systolic ejection murmur in the pulmonary focus.

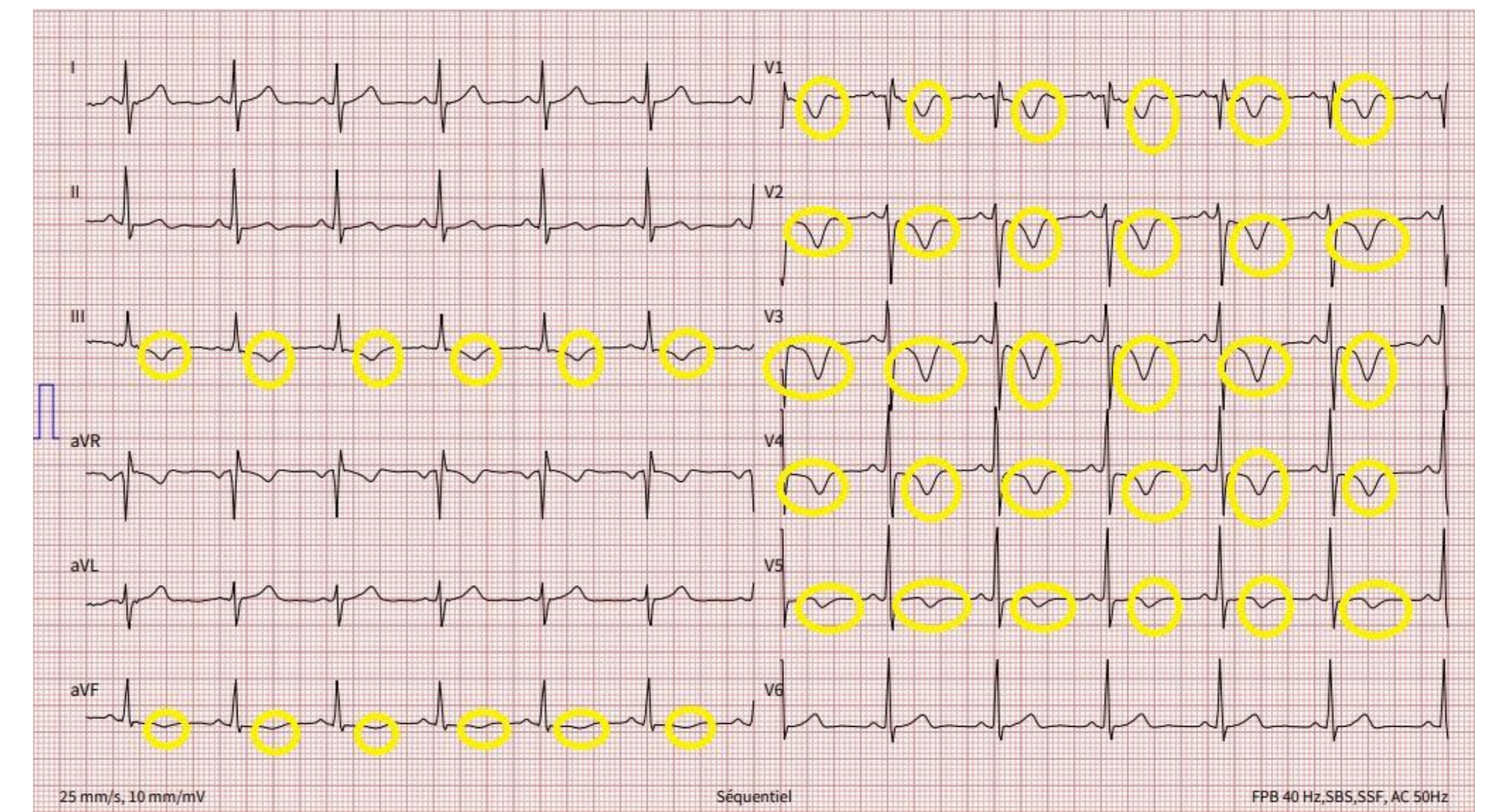


Fig. 1: ECG shows depolarization abnormalities with T-wave inversions in the anteroseptal (V1–V5) and inferior leads (III and aVF), suggestive of ischemia.

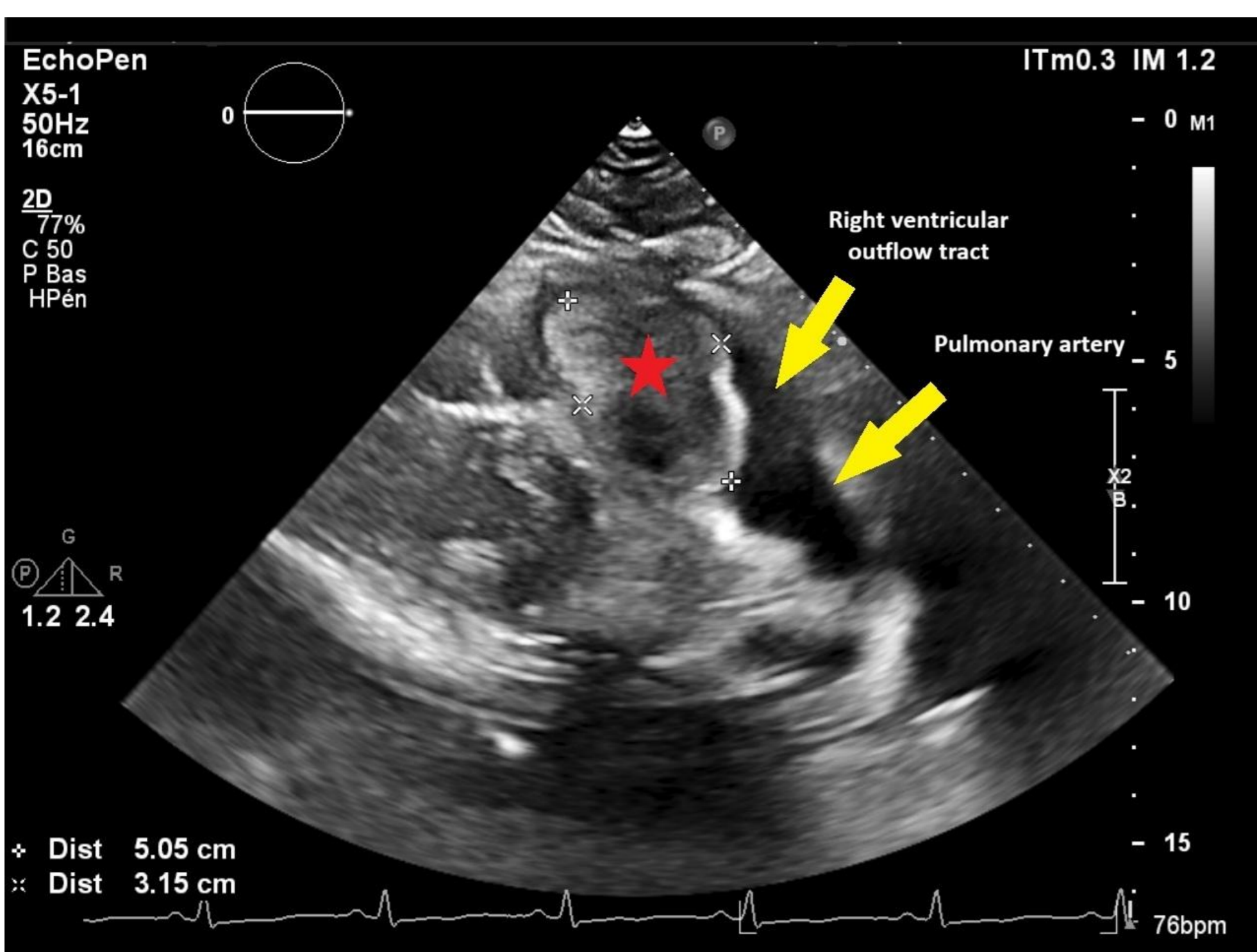


Fig. 2: 2D TTE, parasternal long-axis view of the right ventricular outflow tract, demonstrates a 50 × 31 mm hyperechogenic mass (red star)

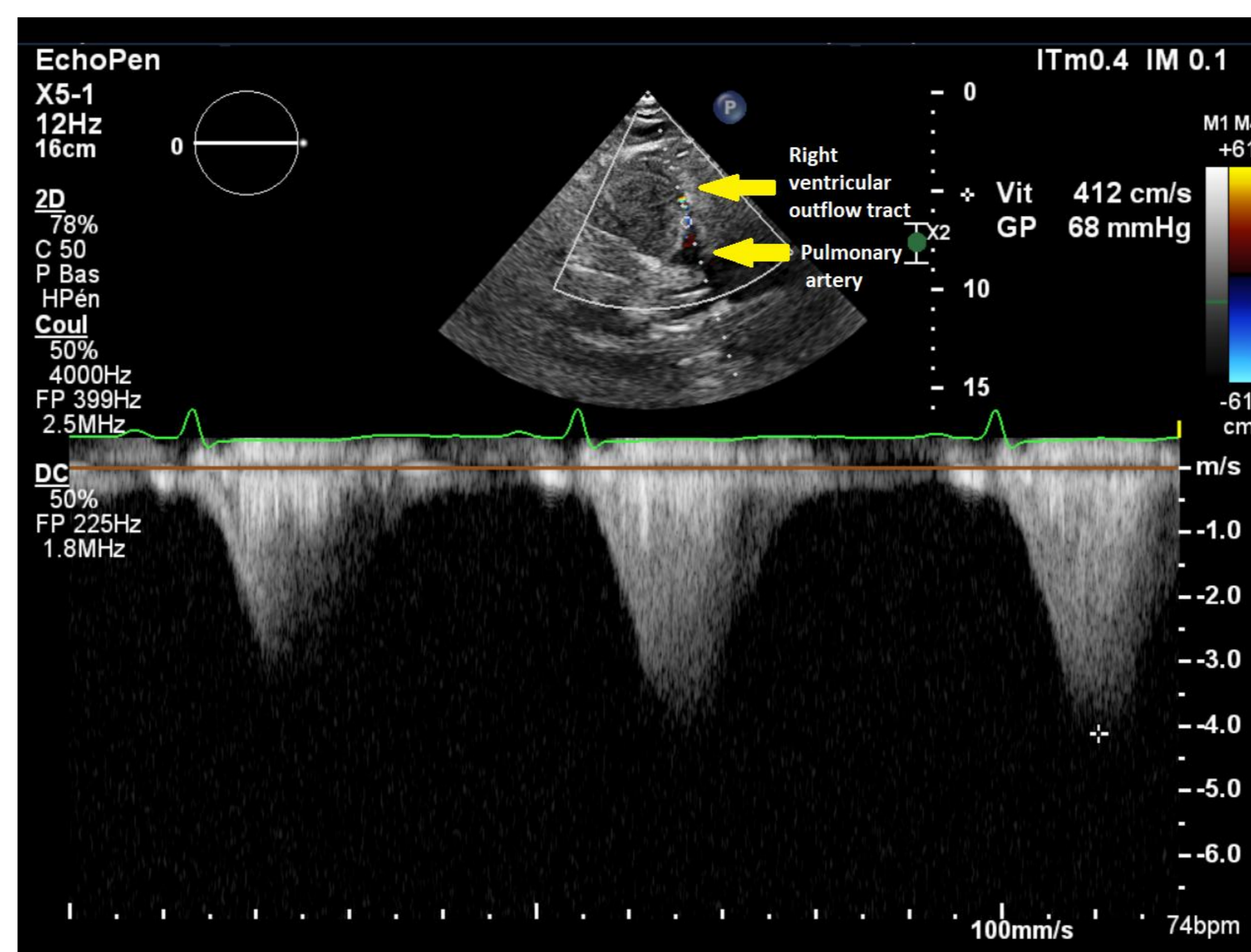


Fig. 3: 2D TTE, parasternal long-axis view of the right ventricular outflow tract, continuous wave spectral Doppler, demonstrates that the mass creates significant obstruction of the outflow tract with a peak systolic pressure gradient of 68 mmHg.



Fig. 4: Thoracic CT scan confirmed a 35 x 53 mm mass, attached via a pedicle to the right ventricle wall.

Primary Cardiac Tumors

- Rare, prevalence of 0.002–0.3%
- ≈ 75% are Benign
- Of these, about ≈ 5% Hemangiomas

Clinical Presentation of Cardiac Hemangiomas

- Mainly asymptomatic
- Valvular obstruction
- Myocardial ischemia
- Heart failure
- Pericardial effusion
- Tamponade
- Systemic embolism
- Conduction disturbances
- Sudden death

Prognosis

- Excellent after surgical removal
- Low risk of recurrence

The adolescent underwent cardiac surgery with resection of the mass. Histopathology revealed a capillary hemangioma.

At one-month postoperative follow-up, the patient was asymptomatic. Physical examination, ECG, and echo were all within normal limits.

The child with chest pain

- Exertional
- Associated with palpitations or syncope
- History of congenital heart disease
- Family history of sudden death or inherited cardiac disease
- Abnormal clinical exam (ie new murmur)
- Abnormal ECG

Take home message

A thorough medical history and physical examination enable the pediatrician to promptly refer the patient to pediatric cardiology, preventing complications and allowing rapid recovery after surgery.

Fig.5: image generated by AI

Intestinal complications from delayed magnet ingestion diagnosis in a paediatric patient with suspected Pica

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Case presentation

- ✓ 11-year-old female
- ✓ History of ADHD, suspected Pica and untreated chronic constipation
- ✓ 48h of diffuse acute abdominal pain and food-relating vomiting
- ✓ No foreign body (FB) ingestion in history

Clinical evolution

- ✓ Fever
- ✓ Migration of abdominal pain in the right iliac fossa
- ✓ Bilious vomiting, progressively acute abdomen

Initial exams

- ✓ Leukocytosis **14.0 G/L**, **CRP 127 mg/L**, with a maximum CRP of **183mg/L**
- ✓ No evidence of appendicitis, intussusception, or free peritoneal fluid on initial abdominal ultrasounds

Medical interventions

- ✓ Broad spectrum IV antibiotics for early peritonitis
- ✓ Enemas and oral laxatives, ineffective
- ✓ Ano-retro-colonoscopy, without viewing FB



Figure 1
Abdominal CT scan after 24h of hospitalization reveals 6 metallic magnetized FB, initially suspected to be in the sigmoid colon, with no signs of perforation



Figure 2
Abdominal Xray initially showing the FB in the sigmoid colon. No signs of major pneumoperitoneum

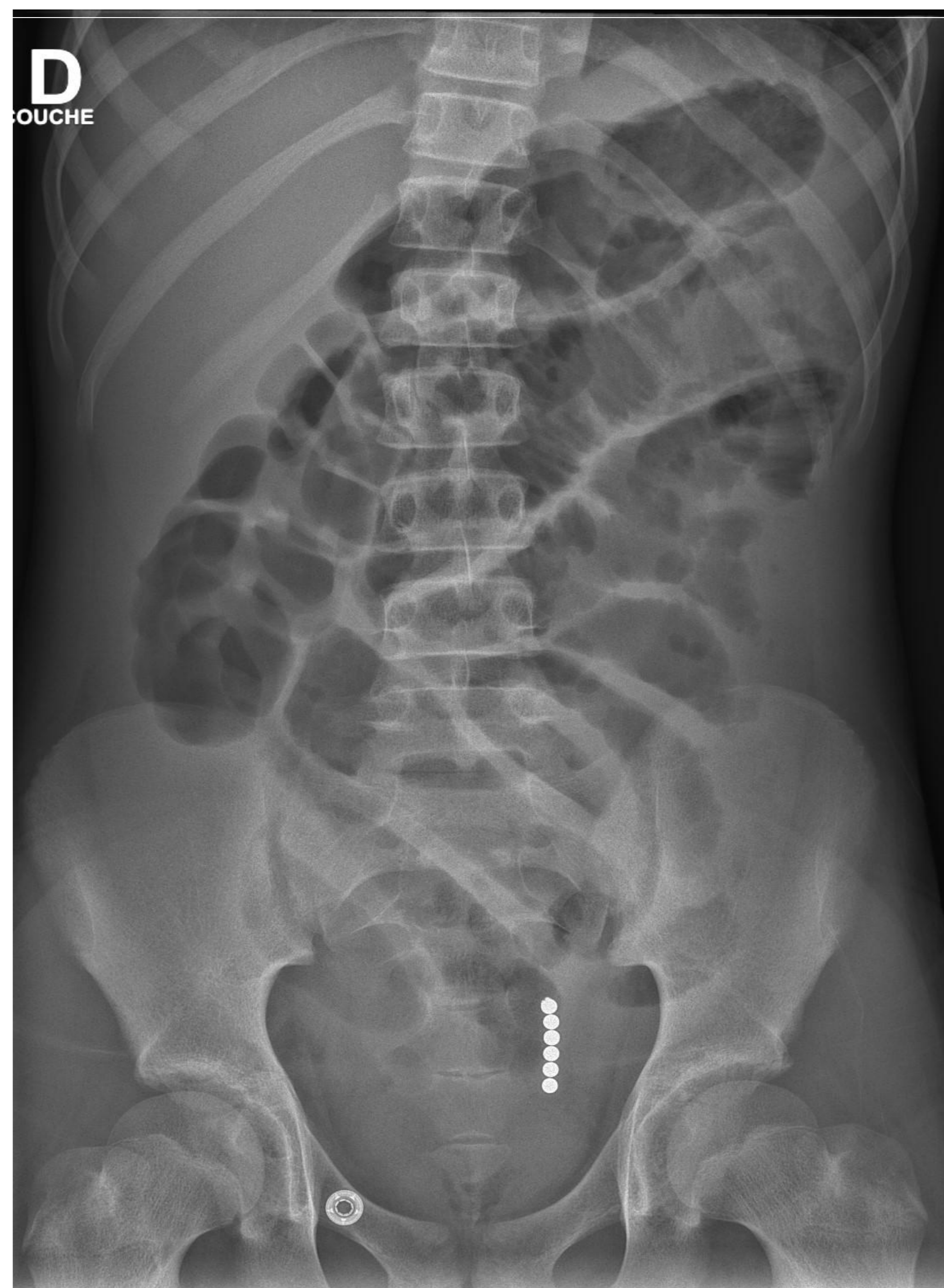


Figure 3
Abdominal Xray after colonoscopy shows stability of FB position, projected in the left lower pelvis

Surgical intervention

- ✓ Exploratory laparotomy on day 5 with successful FB extraction, located outside the intestines
- ✓ Post-operative course uneventful

In this case, the patient gradually developed secondary ileus and peritonitis following a previous intestinal perforation

Discussion

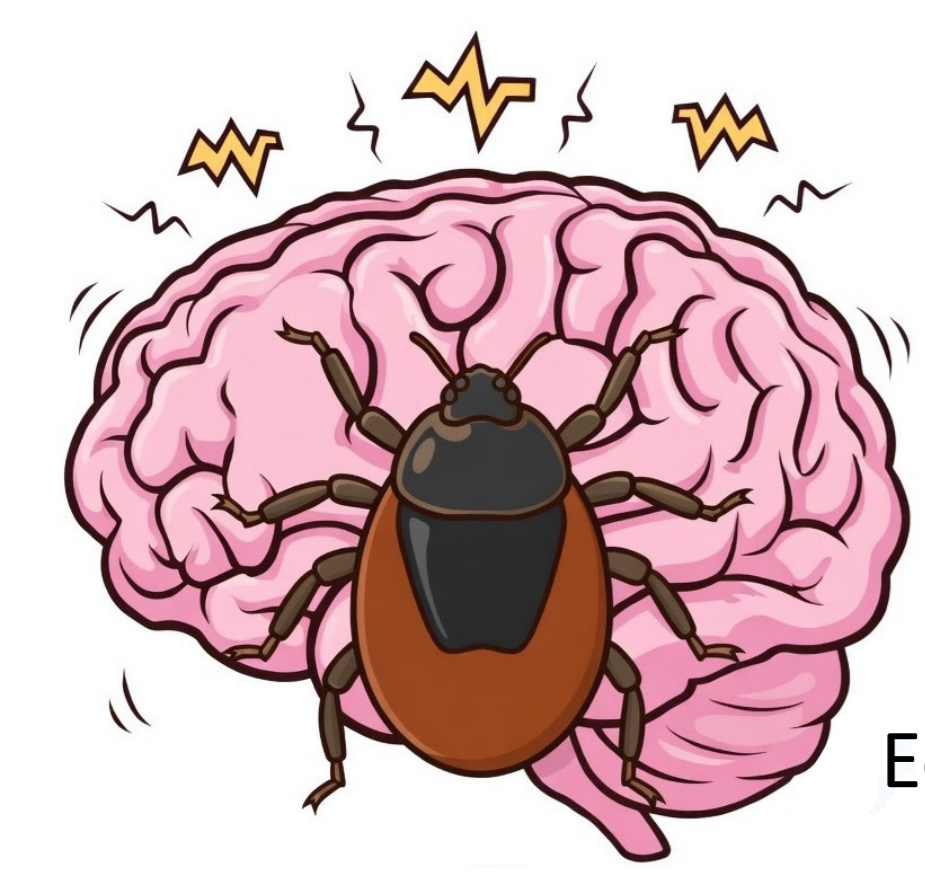
Ingestion of magnetized metallic beads is a risk factor for intestinal complications

Pedopsychiatric comorbidities, such as ASD, personality disorders, depression and eating disorders are risk factors

Reassessing medical history is crucial in the setting of worsening symptoms or failure to improve

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Transient Ischemic Attacks Revealing Neuroborreliosis

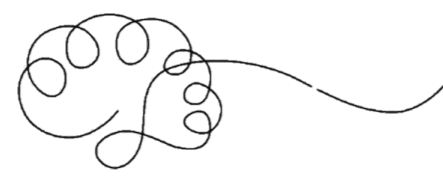


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INTRODUCTION



Pediatric neuroborreliosis is a well-established manifestation of Lyme disease, most commonly presenting with facial nerve palsy or meningitis. Cerebral vasculitis, though very rare, has been described as a complication of neuroborreliosis. We report the case of an adolescent with cerebrovascular involvement, emphasizing the importance of early recognition in endemic areas. (1)

CLINICAL PRESENTATION

A 14-year-old adolescent girl with a history of treated essential hypertension presented with a 3-month history of daily **right-sided headaches**, sometimes nocturnal, associated with **neck pain, vertigo, and nausea**. She was admitted for worsening symptoms with **intermittent focal neurological deficits**, including left-sided paresthesia, transient limb weakness, and possible dysarthria. Neurological examination was normal between episodes. Migraine with aura was initially suspected.

RADIOLOGICAL WORK UP

Brain Angio MRI was performed and revealed **multifocal intracranial vasculopathy** predominantly involving the **posterior** circulation (figure 1).

LABORATORY WORK UP

Cerebrospinal fluid (CSF) analysis showed **lymphocytic pleocytosis** with elevated protein and low glucose. Serological and CSF studies confirmed **neuroborreliosis**, with **positive Lyme serology** (IgG CLIA and Immunoblot), **intrathecal anti-*Borrelia burgdorferi* antibody synthesis** and **positive Lyme CSF PCR**. Chemokine CXCL13 returned also positive.

DISCUSSION

Given the transient nature of neurological deficits and supportive radiological findings, these episodes were considered **transient ischemic attacks**. Combined with microbiological work up, a diagnosis of **neuroborreliosis** with **meningitis** and **cerebral vasculitis** was established.

MANAGEMENT

Intravenous **ceftriaxone** was administered for four weeks. **Antiplatelet therapy** with aspirin 100 mg per day was indicated for two years.

FOLLOW UP

After treatment, clinical outcome was favorable, with **complete resolution** of neurological symptoms. Follow-up MRI demonstrated marked regression of vascular abnormalities without ischemic sequelae (figure 2).

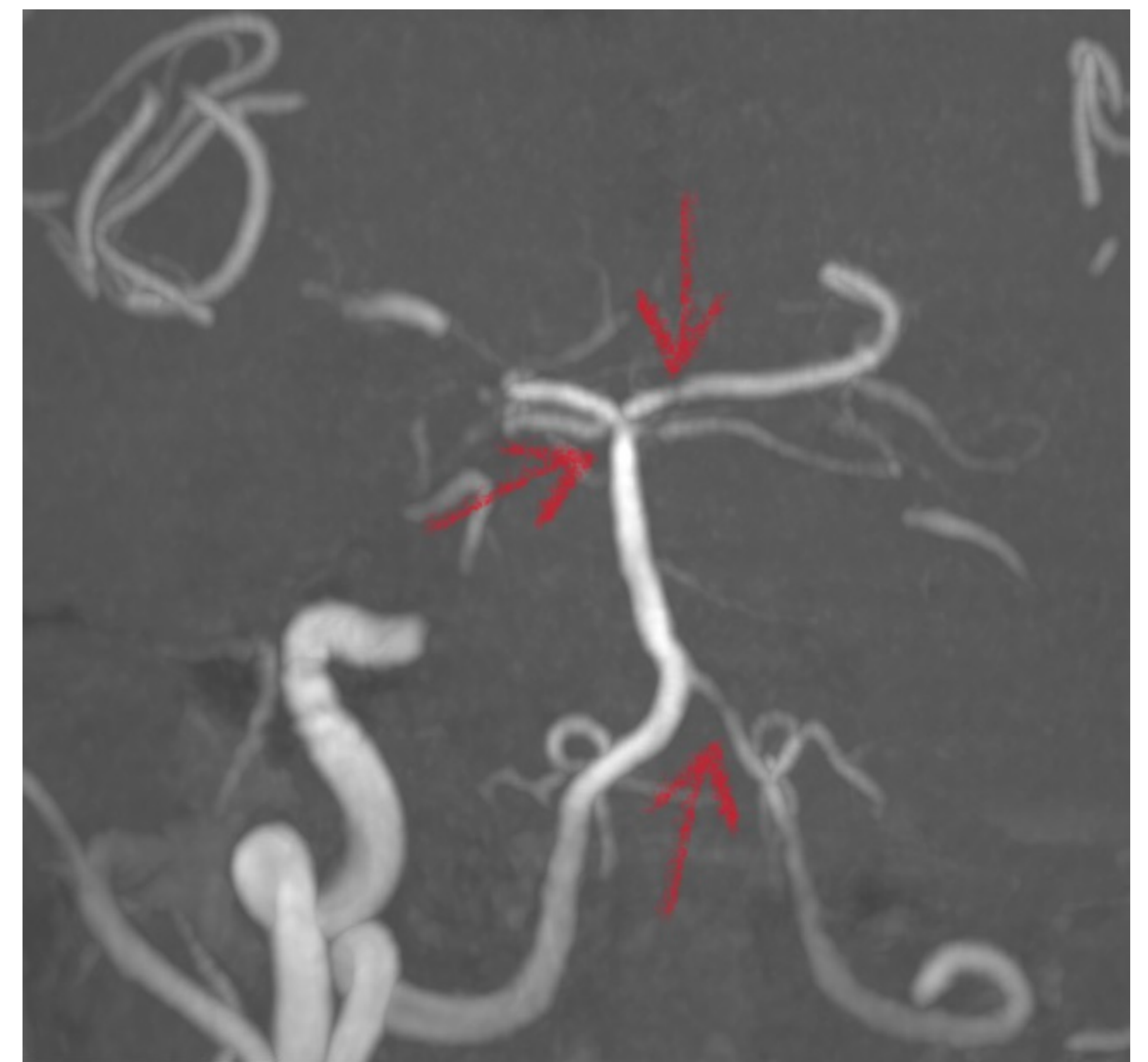


Figure 1. Proximal segment of the left posterior cerebral artery, terminal portion of the basilar artery, and V4 segment of the left vertebral artery (from top to bottom).

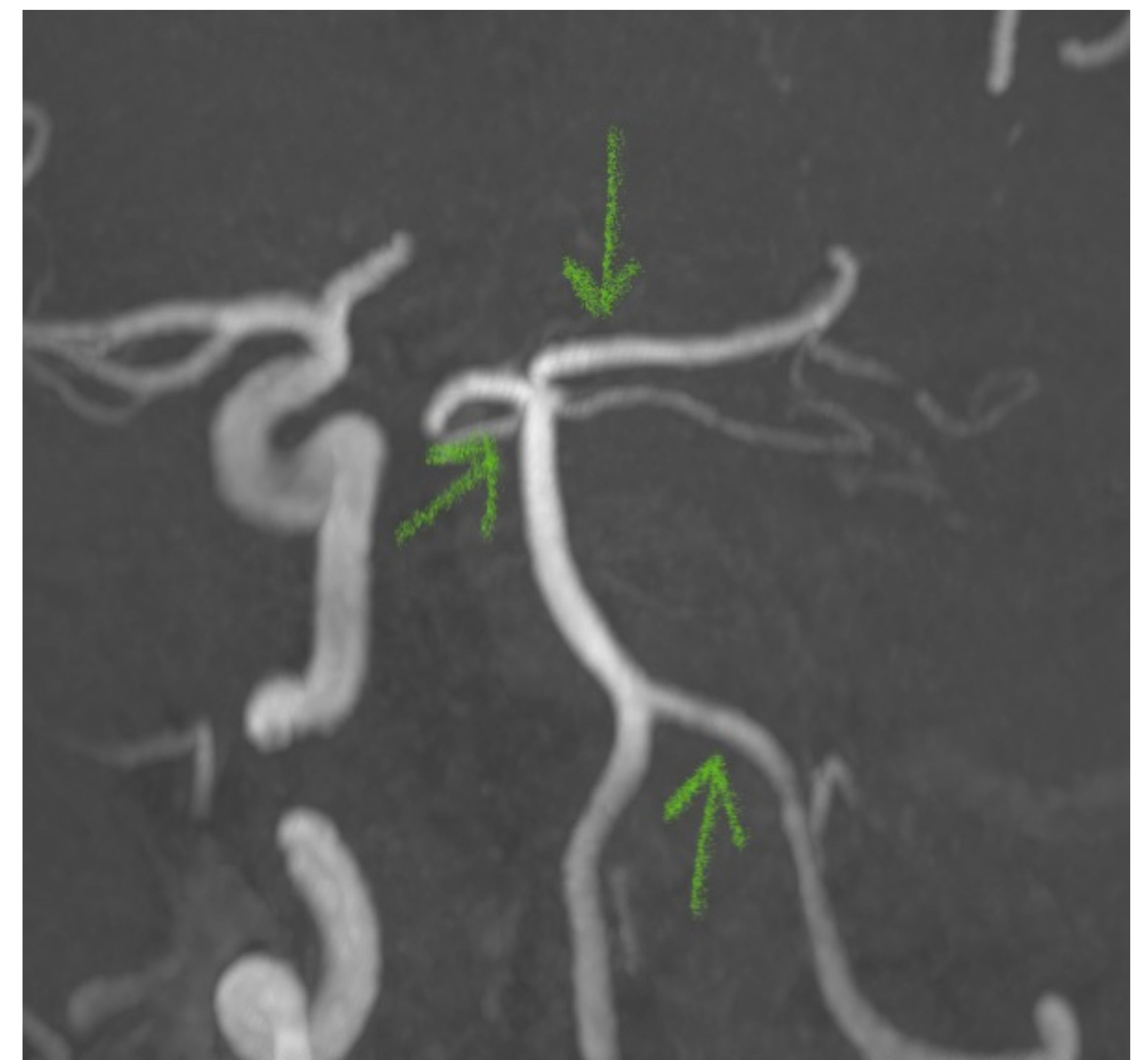


Figure 2. Follow-up MRI obtained 3 months after treatment, showing resolution of the lesions depicted in Figure 1.

CONCLUSION

Neuroborreliosis-associated vasculitis in children is rare but typically presents with **focal neurological deficits and stroke-like symptoms**, with imaging showing **multifocal ischemic infarcts and arterial stenoses**. Diagnosis relies on **intrathecal anti-*Borrelia* antibody production** with **lymphocytic pleocytosis** and exclusion of other pediatric stroke etiologies. Prompt intravenous **ceftriaxone** is recommended and is associated with a favorable prognosis. Complicated neuroborreliosis should be considered in any **unexplained stroke or transient ischemic attack in endemic areas**. (2,3)

Herniated discs: when pediatrics meets geriatrics - a case report

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Introduction

Lower back pain (LBP) is a common complaint among pediatric patients, with a prevalence ranging from 20 to 50% and increasing with age [5, 8]. While non-specific musculoskeletal pain is the most frequent cause [10, 12], clinicians must remain vigilant for history and clinical red flags (**Table 1**) suggesting traumatic, infectious, neoplastic, or inflammatory etiologies that should prompt further investigations [4,8,13].

Lumbar disc herniation (LDH) is an uncommon etiology of specific back pain in children. Its incidence increases with age, and trauma is the most associated factor along with positive family history of back pain or herniated disc [2,3, 9]. Clinical features do not differ from the adult presentation with typical radicular pain and symptoms, worsened by trunk flexion and a positive straight-leg test [12].

Case description

A 14-year-old male patient presented to the emergency ward with a two-week history of worsening pain in his left lower limb. At first, the patient did not recall any history of trauma. The pain was located on the lateral side of his left leg and lower back, was worsened by movements, accompanied by paresthesia in the foot, compatible with L4-L5 radicular pain (**Figure 1**). The pain was also present at night. Familial history was positive for Crohn's disease and herniated disc in the patient's father.

Clinical examination revealed a positive straight leg test without neurological deficits. Laboratory results showed no evidence of inflammation or hematologic malignancy, and X-ray of the spine and lower limbs excluded fracture (**Figure 2**). The orthopedic team recommended symptomatic treatment without further evaluation.

The patient returned to the emergency department one week later with worsening pain. He then remembered a fall from a climbing wall a couple of days prior to the pain. Repeated examination revealed a sensorimotor deficit in the L5 dermatome of the left foot, prompting a lumbar MRI, which confirmed a herniated disc at the L4-L5 level (**Figure 3**). After consultation with the neurosurgical team, conservative management was continued. Two weeks later, due to worsening pain and difficulty walking, the patient was admitted for a lumbar microdiscectomy, which led to rapid recovery.

History	Neurological	Systemic	Structural / Other
Age < 10 years	Motor and/or sensory disturbance	Fever	Palpable step deformity
History of Trauma	Radicular / irritative pain	Local swelling	Structural change of the spine
Pain > 4 weeks	Positive straight leg raise test	Lymph node enlargement	Joint hypermobility
Night pain	Bladder or bowel dysfunction	Other B-symptoms	Pre-existing medical conditions
Morning stiffness			

Table 1 – Red flags for back pain in children, adapted from [5, 11, 14].

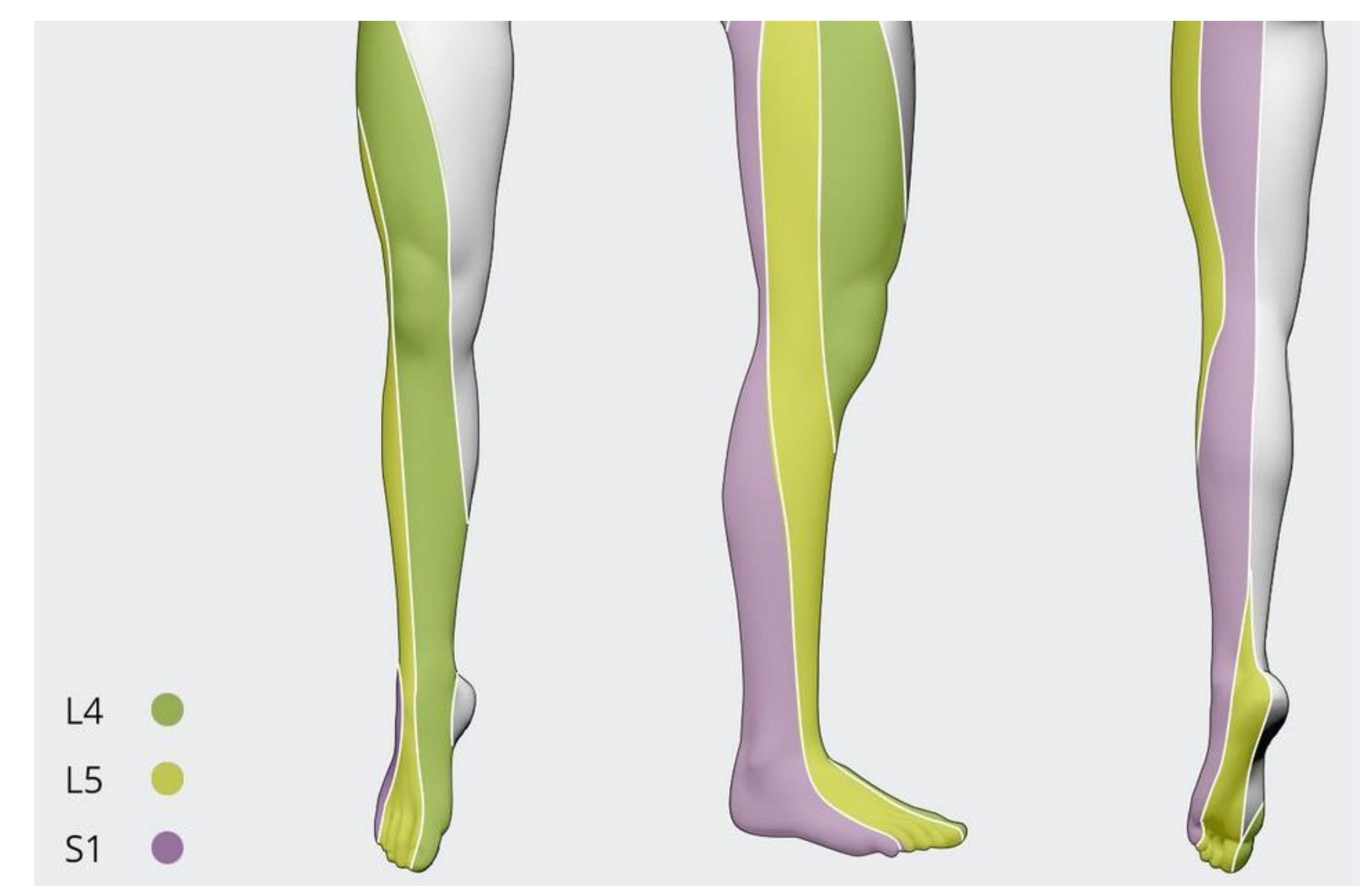


Figure 1 – L4 to S1 dermatome, copyright Neurochirurgie, Inselspital Bern

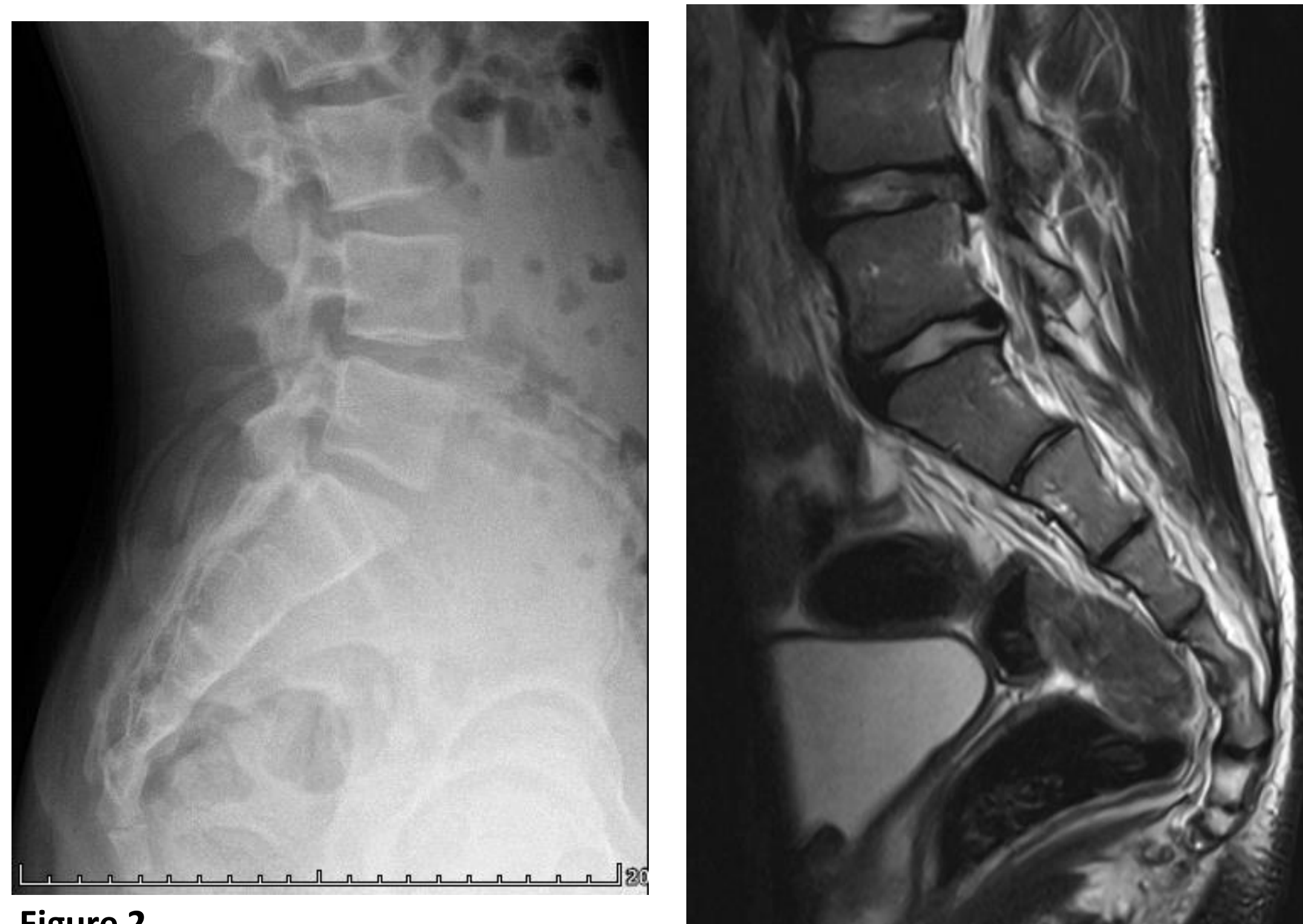


Figure 2
Profile Lumbar X-ray of the patient

T2-sequence spinal MRI of the patient
confirming the L4-L5 herniated disc

Discussion

This case highlights the importance of recognizing red flags in adolescents presenting with low back pain. Indeed, this patient presented with lower back pain, five associated red flags (**Table 1**), several risk factors [2,3, 6, 9], and highly suggestive clinical of LDH [12].

Although current guidelines recommend X-rays as first-line imaging in children with red flags, their diagnostic yield for disc herniation is limited. In the presence of neurological symptoms, early MRI may be more appropriate to avoid diagnostic delay [1].

L4-L5 and L5-S1 are the most affected levels [6, 9, 11], which was also the case in our patient. Initial management is conservative, although less effective in pediatric patients with long-term success of 25-50% [3,9]. Operative treatment is indicated for persistent or severe symptoms, failure of conservative treatment or neurological deficits, with preference for microdiscectomy or endoscopic discectomy [2, 9, 11]. Microdiscectomy is associated with over 80% good immediate outcomes and low complication rates [9]. In our case, the patient experienced good clinical outcome after surgery with total resolution of symptoms at the 2-month follow-up.

This case emphasizes that, although rare, lumbar disc herniation should be considered in adolescents with radicular pain, and that early MRI may be warranted in the presence of neurological symptoms to prevent diagnostic delay and optimize outcomes. Pediatric disc herniation may differ from adult forms in terms of response to conservative treatment and early surgery may improve outcomes.

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An uncommon cause of paediatric lymphadenitis: Mandibular osteomyelitis due to *Actinomyces naeslundii*

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Background

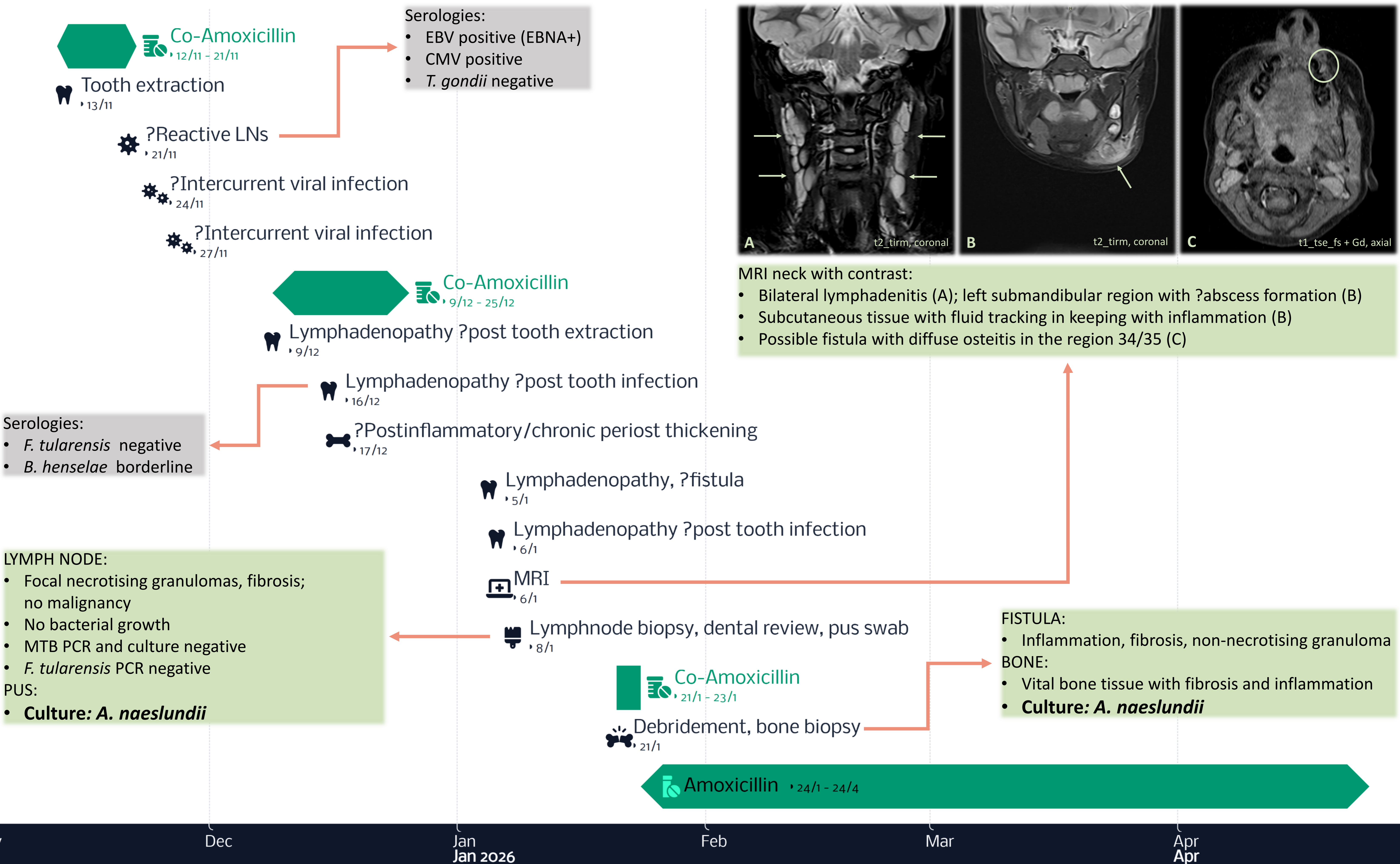
- Cervical lymphadenitis is a common presentation in paediatrics
- Important clues in the history and clinical presentation include:
 - Acute vs. chronic
 - Unilateral vs. bilateral
 - Local findings e.g. inflammation
 - Systemic signs and symptoms
- Differential diagnoses include infections, inflammatory conditions and malignancy

The Case

- 7-year-old female
- Presentation with persistent left submandibular lymph node enlargement first appearing two weeks after a left molar extraction due to caries
- Mostly afebrile, systemically well
- Previously well, fully vaccinated, no known allergies
- No relevant travel or exposure history

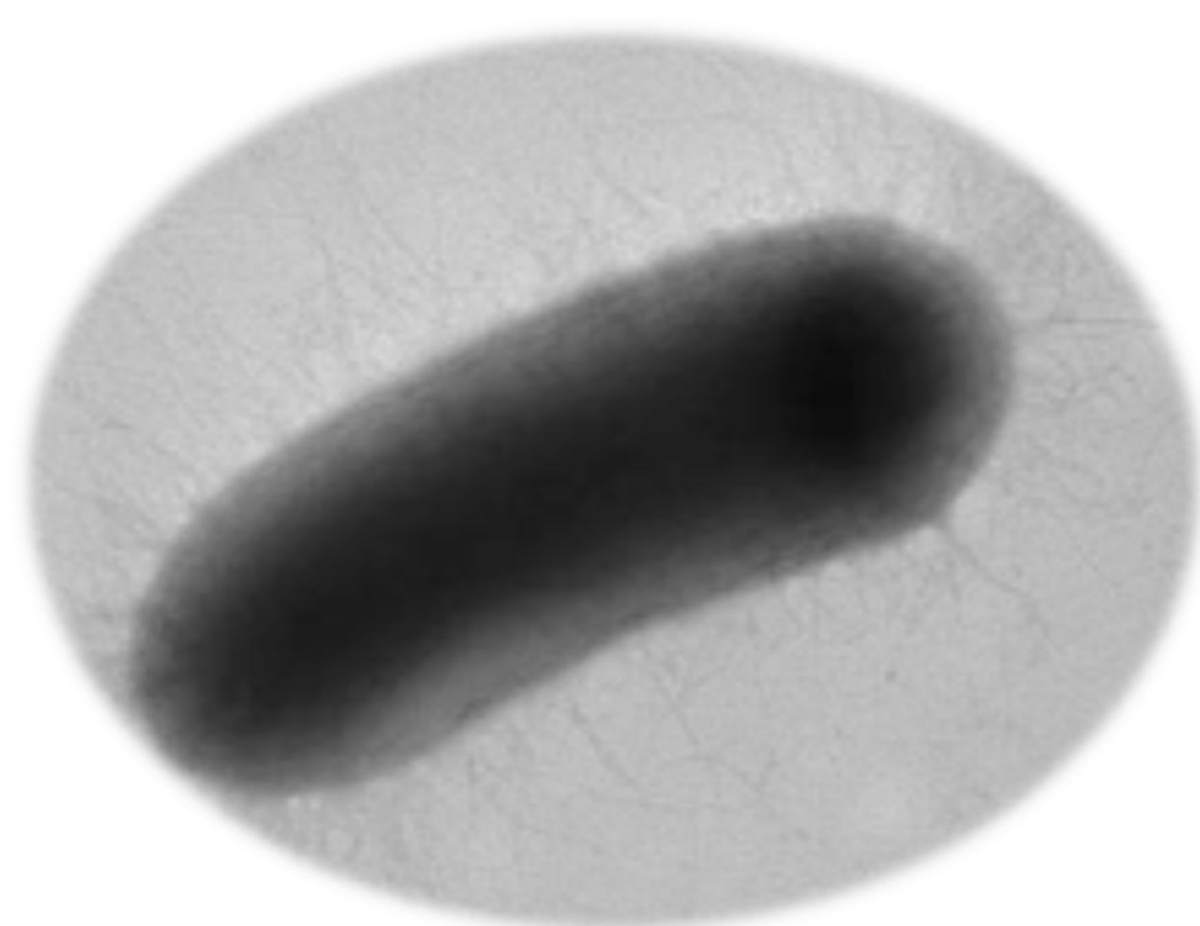


The timeline



Actinomyces

- Common oral, gastrointestinal and respiratory commensals
- Rarely cause infections in immunocompetent children
- Slow-growing, gram-positive branched rods
- Pathogen introduction following breakdown of mucocutaneous barriers
- Spread by direct invasion of adjacent tissues
- Formation of sinus tracts common
- Three common anatomical sites of infection
 - Cervicofacial "lumpy jaw" (after tooth extraction/oral surgery)
 - Thoracic (aspiration/extension of cervicofacial infection)
 - Abdominal (after penetrating trauma/intestinal perforation)
- Treatment with high dose penicillin, prolonged treatment for osteomyelitis



Conclusions

- Horses before zebras, but don't forget the stripes if things don't add up
- Tonsils and the pharynx are important, but in front is a whole oral cavity
- Samples count; consider when, where, what

Quand une habitude courante entraîne une complication inhabituelle Panaris compliqué d'ostéomyélite

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Introduction

Nous rapportons le cas d'une adolescente de 12 ans présentant une ostéomyélite de la phalange distale causée par un panaris d'évolution défavorable dans un contexte d'onychophagie chronique.

Le panaris est une infection fréquente en pédiatrie, généralement bénigne. Dans de rares cas, une évolution défavorable peut mener à des complications ostéo-articulaires nécessitant une prise en charge chirurgicale urgente.

Case Report et Évolution Clinique

- **J0**
 - Panaris (D2 main droite) évoluant depuis 2 jours
 - Consultation dans un centre médical : drainage + désinfection
 - Traitement à domicile: ATB PO : co-amoxicilline 25mg/kg 2x/j pour 7 jours

- **J7**
 - Péjoration des douleurs
 - Nouvelle consultation : débridement nécrose de la pulpe
 - Changement d'ATB : cefuroxime 500mg 2x/j

- **J14**
 - Consultation aux urgences pédiatriques (SAUP)
 - Nécrose de la pulpe avec exposition osseuse
 - Bilan biologique normal (FSC et CRP)
 - RX D2 F/P : ostéomyélite phalange distale
 - Avis chirurgical demandé



- **J15 (J0 post-op)**
 - Débridement chirurgical
 - Biopsie osseuse + frottis profond pour culture
 - Co-amoxicilline IV 35mg/kg 3x/j pendant 3 jours



- **J18 (J3 post-op)**
 - MSSA identifié sur culture osseuse et frottis profond
 - Amélioration clinique relais AB par
 - Co-amoxicilline PO 30mg/kg 3x/j pour 4 semaines



- **J27 (J6 post-op)**
 - Cicatrisation en cours



Discussion

Fréquence et gravité :

- Infection fréquente chez l'enfant
- Le plus souvent bénigne
- Complications rares

Facteurs de risque :

- Onychophagie chronique, succion des doigts
- Diabète type 2, immunosuppression, ø traitement

Évolution inhabituelle :

- Aggravation malgré traitement initial = anormal
- Suspecter une infection profonde

Démarche diagnostique :

- Réévaluation clinique indispensable
- Imagerie si évolution défavorable (RX ou IRM)
- Rechercher une atteinte osseuse

Prise en charge :

- Formes compliquées = prise en charge multidisciplinaire
- Débridement chirurgical
- Antibiothérapie IV adaptée

Épidémiologie¹ panaris compliqués

Population

- 125 patients opérés sur 16 mois

Étiologies

- Plaies : 23 cas
- Piqûres végétales : 21 cas
- Cause inconnue : 25 cas

Complications : 33 % (41 patients)*

- Ostéomyélites : 16
- Ostéoarthrites : 6
- Phlegmons des gaines : 15
- Extensions infectieuses : 6
- Formes nécrosantes : 5

Délai de prise en charge 1^{er} consultation

- 18 jours en moyenne
- 12 jours (formes non compliquées)
- 30 jours (formes compliquées)

*Certains patients présentaient plusieurs complications

Conclusion

- Réévaluation nécessaire dès évolution défavorable
- Prise en charge multidisciplinaire
- Evite des séquelles esthétiques et fonctionnelles

Références

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Pott's Puffy Tumor Complicating Frontal Sinusitis in an Adolescent: Case report

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HIB

HÔPITAL
INTERCANTONAL
DE LA BROYE

BACKGROUND

- Pott's puffy tumor (PPT) is a rare but potentially life-threatening complication of frontal sinusitis.
- Osteomyelitis of the frontal bone with subperiosteal abscess.
- Most common in adolescents.
- Risk of intracranial complications (epidural abscess, cerebritis, venous thrombosis).

CASE

- 13-year-old boy, previously healthy
- Day 0: frontal sinusitis for 10 days → oral antibiotics (amoxicillin-clavulanate)
- Day 14: midline frontal swelling + persistent headache
- No fever, no neurological deficits
- Leucocytosis, elevated inflammatory markers
- CT: frontal sinusitis + epidural abscess + subcutaneous collection (Pott's puffy tumor)

MANAGEMENT

- IV ceftriaxone (meningitic dose).
- Surgical drainage (Day 14).
- Metronidazole added (anaerobic coverage).
- Culture of surgical drainage material: ***Streptococcus anginosus***.
- IV therapy adjusted according to antibiogram.

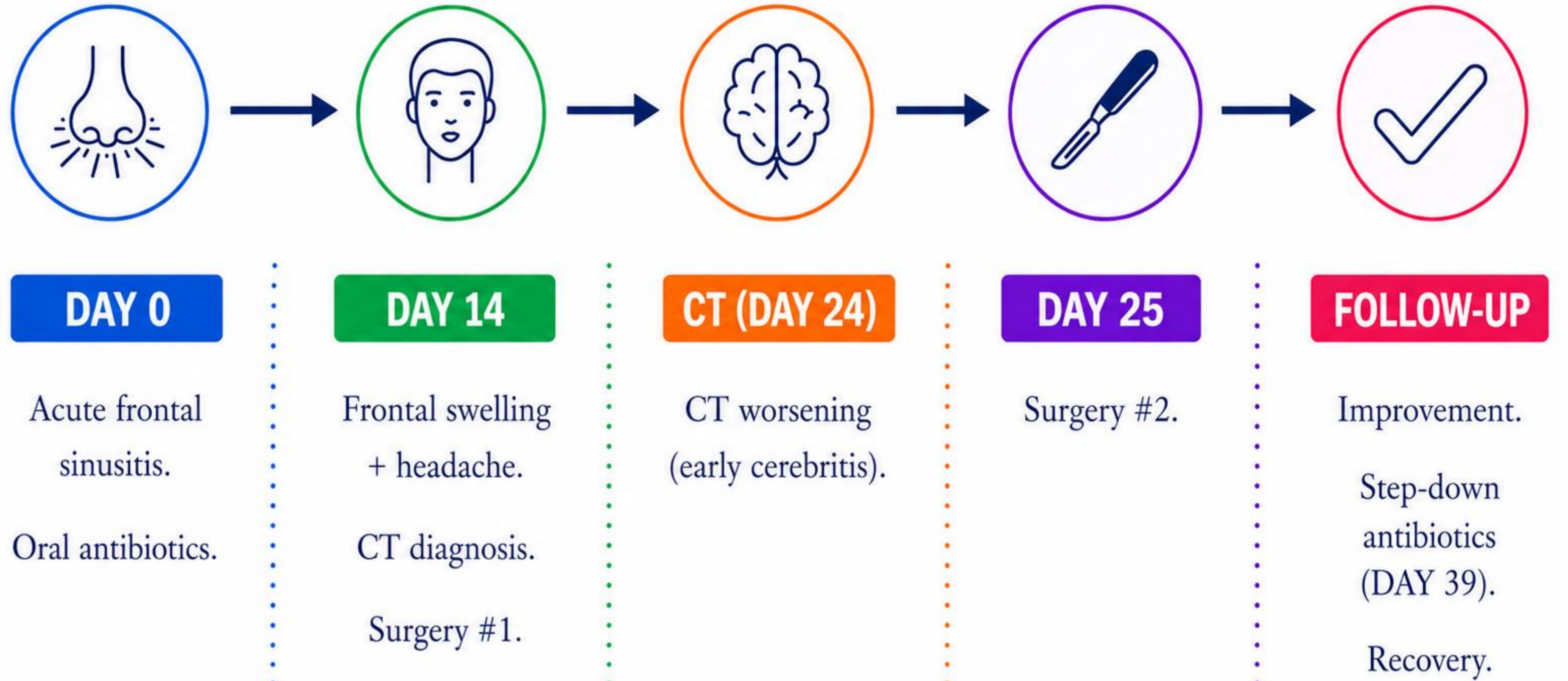
CLINICAL COURSE

- CT Day 24: worsening + early cerebritis.
- Second surgical drainage.
- Clinically stable throughout.
- Serial imaging: improvement.
- Day 39: step-down to oral antibiotics (amoxicillin).
- Discharged in good condition.
- No neurological sequelae.

KEY MESSAGES

- PPT can progress despite appropriate therapy.
- Early imaging and close monitoring are crucial.
- Multidisciplinary management improves outcomes.
- Prolonged targeted antibiotics and surgical drainage are key.

TIMELINE

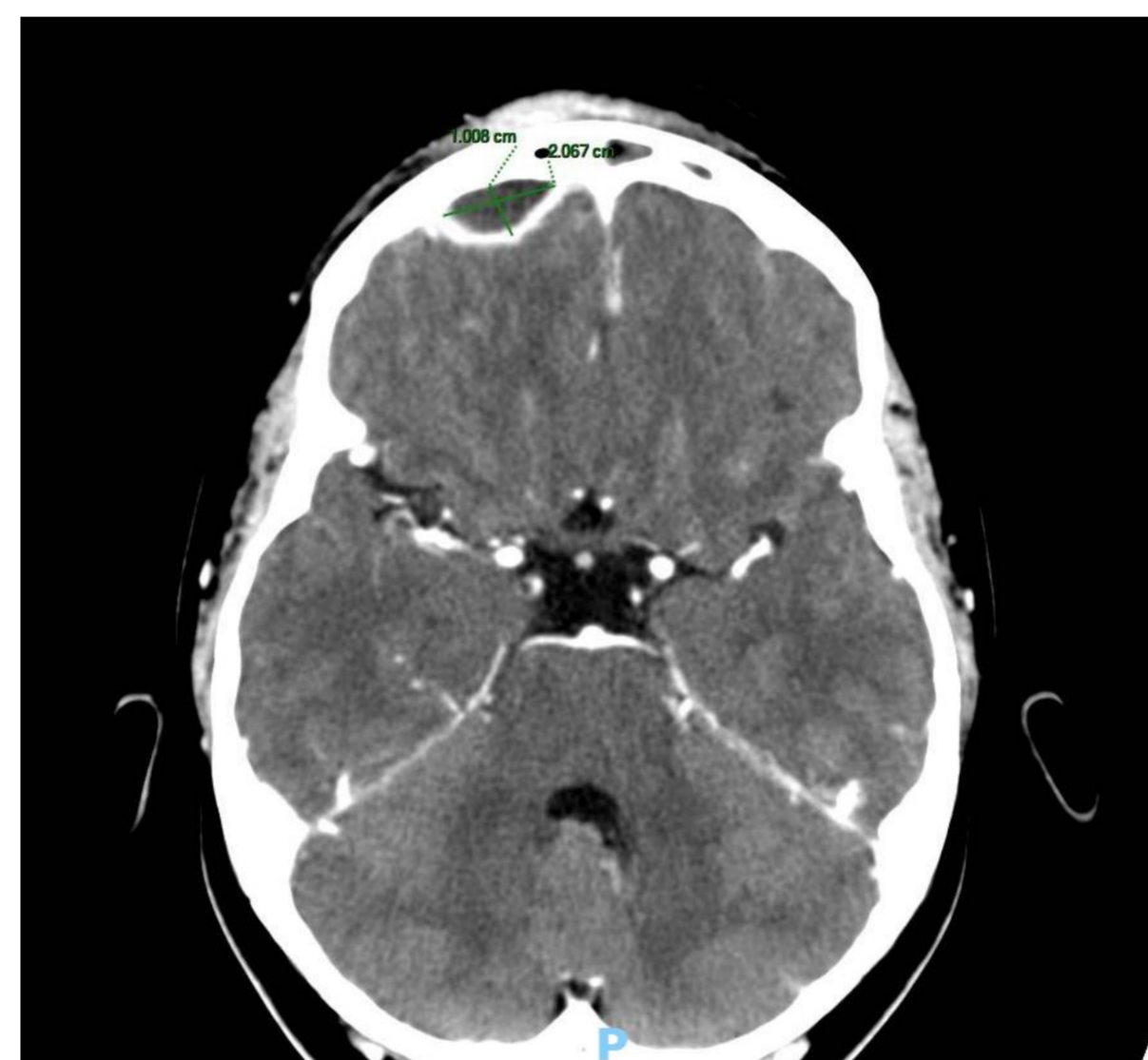


CLINICAL PHOTOS



IMAGING

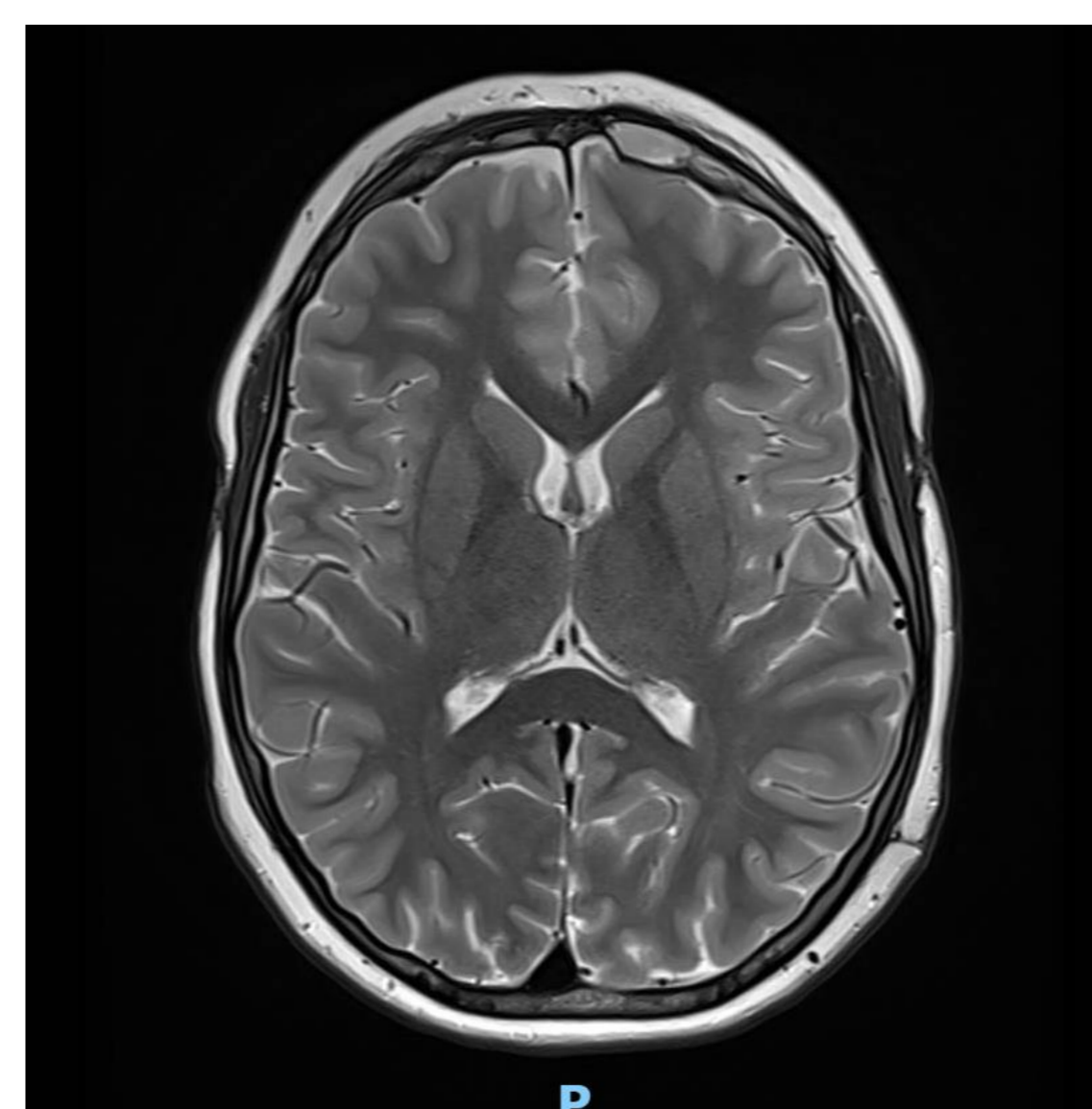
1 INITIAL CT (DAY 14)



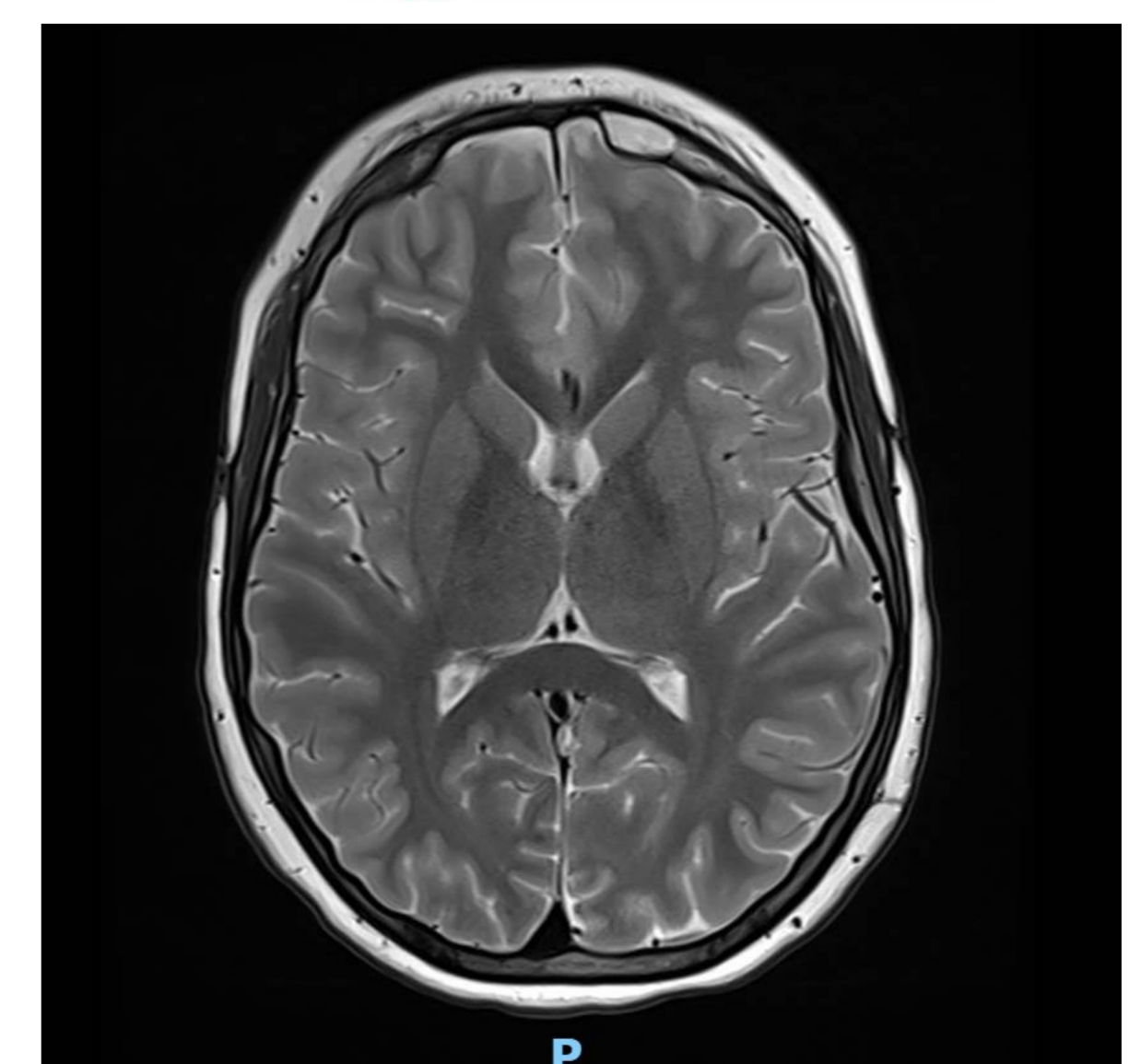
2 CT (DAY 24)



3 MRI (3 MONTHS)



4 MRI (6 MONTHS)



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Background

Dystonia is a movement disorder defined by involuntary and sustained contraction of muscles leading to abnormal movements or posture. It is associated with perturbations in sleep, rest, and daily activities, but its recognition and management remain challenging. In pediatricians' consults, dystonia is mainly encountered in patients with cerebral palsy, metabolic diseases, or neurological disorders. Proactive identification and prevention of dystonia triggers can be done by **pediatricians in outpatient setting**, and **coordination of interprofessional care** is essential to improve patient quality of life.

Case report

A 5 year-old girl known for neurodegeneration with brain iron accumulation developed **secondary focal dystonia** in the right lower limb **managed in outpatient setting**.

Following **subacute worsening of dystonia in the context of a viral infection**, she was **addressed by her pediatrician** to the pediatric emergencies and admitted in the pediatrics ward.

A **dystonia care pathway** was initiated:

- **general care**: optimized hydration and sleep
- **physical interventions**: optimized sleeping and seating positioning, environmental adaptations by **occupational therapists** and **physiotherapists**, nutritional management by **dietitians**, swallowing support by **speech therapists**
- **pedopsychiatric support**
- **non-specific pharmacological interventions**: antipyretics, laxatives
- **pro-active pain management**: non-opioid and opioid analgesics
- **dystonia-specific pharmacological intervention**: anticholinergics, benzodiazepines, clonidine.

With **daily reassessment of clinical status by nursing and medical teams**, dystonia was pro-actively addressed to improve patient's clinical evolution and quality of life.

A **deep brain stimulation (DBS) implant** was placed neurosurgically. She later required **admission to the PICU for a severe sepsis on catheter infection** leading to **status dystonicus**.

In the follow-up, she has now **returned to standard care with stable dystonia**.

Central role of the pediatrician in dystonia care pathway:

- Identify and prevent dystonia triggers
- Reevaluate dystonia severity
- Coordinate interprofessional care
- Integrate specialized care such as neuropaediatrics, neurorehabilitation, and neurosurgery

Common dystonia triggers to look for

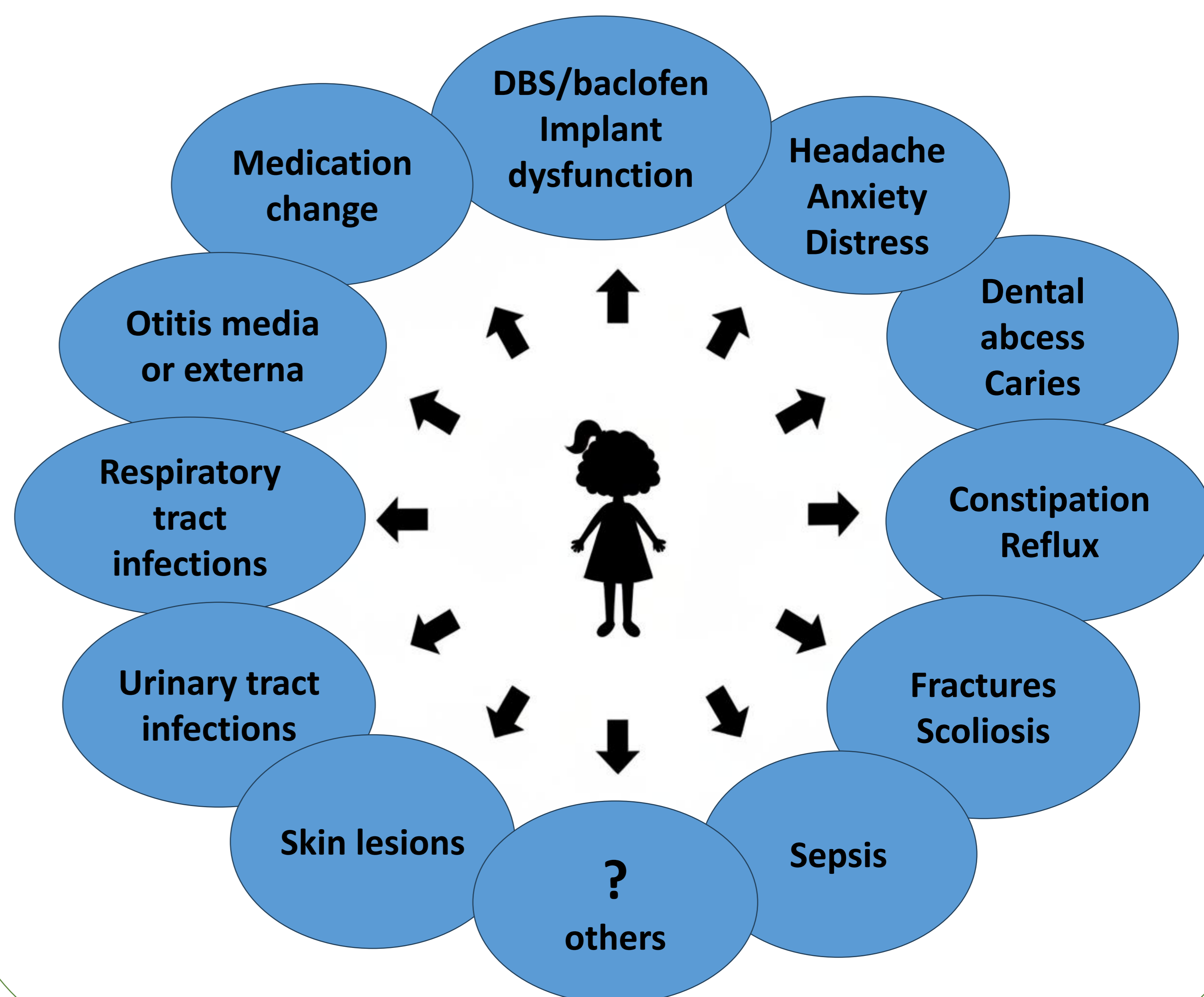


Fig. 1: Dystonia triggers identification requires detailed clinical evaluation (adapted from Lumsden DE, *Arch Dis Child Educ Pract Ed.*, 2025)

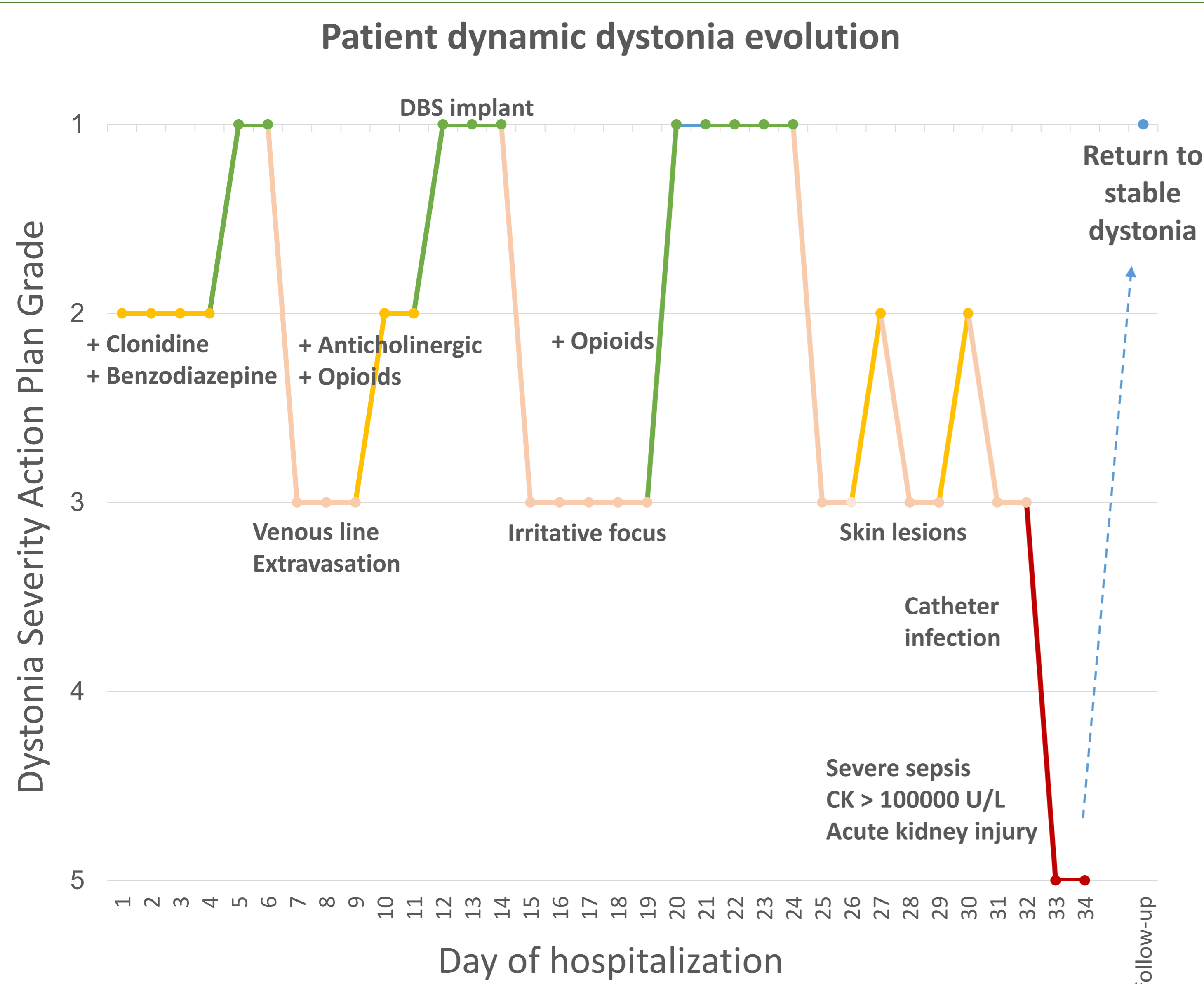


Fig. 2: Patient dystonia severity action action plan grade during her hospitalization.

Dystonia Severity Action Plan Grade	Features	Action
1	Sits comfortably, regular sleep, stable	Nil required
2	Irritable, cannot settle, posturing interfere with seating, only tolerates lying	Urgent outpatient evaluation
3	Cannot tolerate lying, sleep disturbed, no signs of metabolic or airway compromise	Acute admission
4	Clinically grade 3, with metabolic disturbance (e.g CK >1000)	Continuous care management
5	Severe dystonia, metabolic decomp., respiratory or CV compromise requiring	organ support Pediatric intensive care unit management

Fig. 3: Dystonia severity action plan (adapted from Lumsden DE, *Arch Dis Child Educ Pract Ed.*, 2025)

Reference

Lumsden DE. *Archives of Disease in Childhood - Education and Practice* 2025;110:8-14 doi: 10.1136/archdischild-2023-326814

HEMORRHAGIC CYSTITIS AS A RARE MANIFESTATION OF SARS-COV-2 IN A NEONATE: A case report.

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BACKGROUND

SARS-CoV-2 infection is mainly associated with respiratory disease, but extrapulmonary manifestations are increasingly recognized. Hemorrhagic cystitis is a rare clinical presentation [1].

CASE REPORT



Figure 1. Urine sample showing gross hematuria.

A one-month-old male infant **presented with high fever (>39 °C), decreased oral intake, and macroscopic hematuria.** Pregnancy and perinatal history were unremarkable. He was febrile, hemodynamically stable, without respiratory symptoms.

An infectious workup was performed. Urinalysis confirmed gross hematuria and urine culture excluded bacterial UTI. Renal and bladder ultrasonography were normal.

A respiratory PCR panel was positive for SARS-CoV-2. Given the temporal association, negative urine analysis and culture, and no evidence of other etiologies, hemorrhagic cystitis is likely associated with SARS-CoV-2. The patient received supportive care with clinical improvement. On day 3, a secondary UTI developed, treated with antibiotics, possibly related to the prior viral urothelial injury.

DISCUSSION

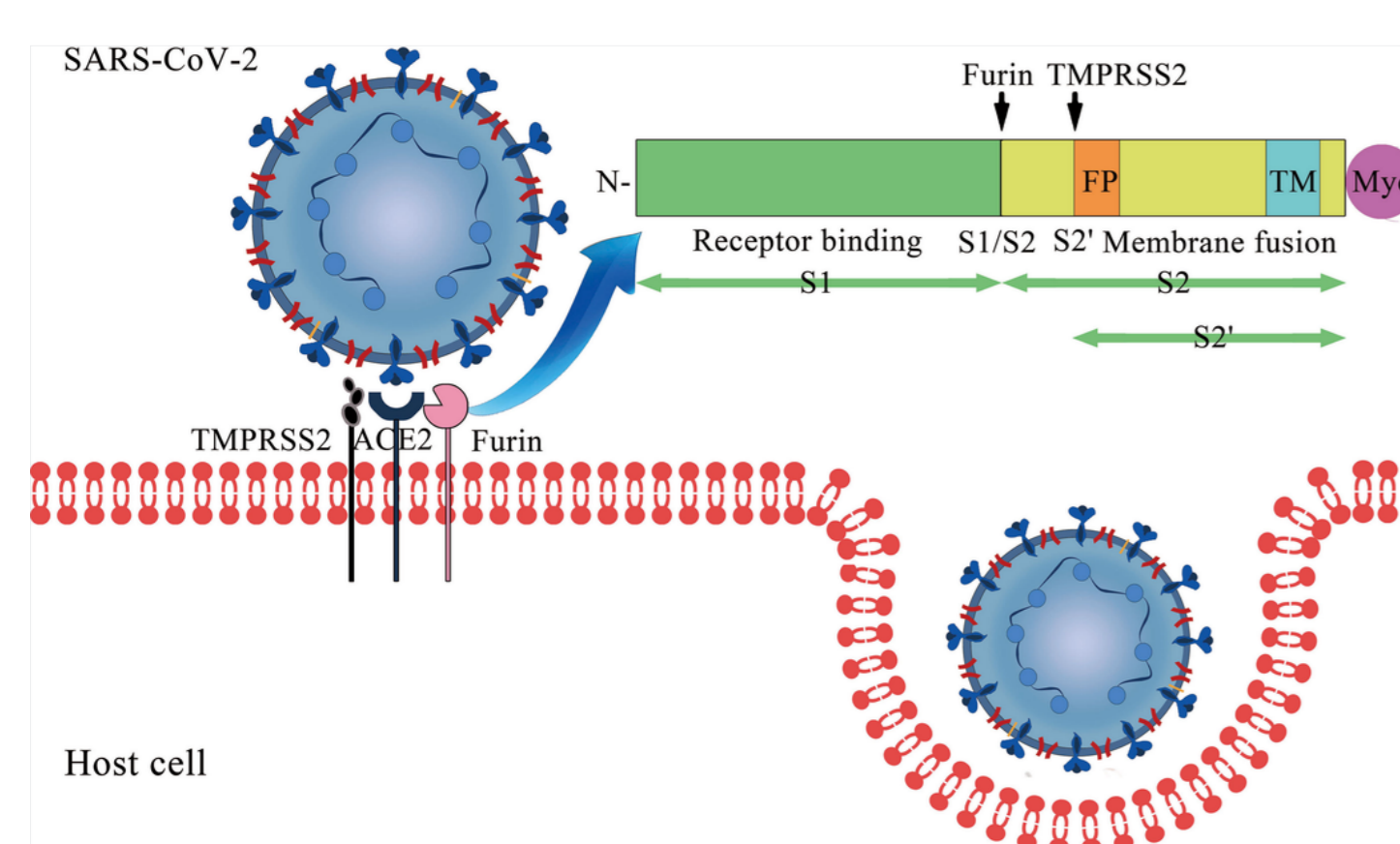


Figure 2. Interaction between the SARS-CoV-2 Spike protein and the membrane ACE2 receptor Chen et al., 2021.

SARS-CoV-2 can affect multiple organs. Hemorrhagic cystitis is rarely described in children and is exceptional in neonates [1]. The pathophysiology is unclear, but the expression of **angiotensin-converting enzyme 2 (ACE2) receptors on urothelial**

cells may allow direct or immune-mediated viral injury. Endothelial dysfunction, inflammation, and cytokine-mediated damage may also contribute [2].

In this case, negative initial urine culture, normal imaging, and the close temporal relationship with SARS-CoV-2 infection support a diagnosis of COVID-19-associated hemorrhagic cystitis. Subsequent UTI suggests viral urothelial damage may predispose to bacterial infection.

URINALYSIS RESULTS

Parameter	Reference range	22 Jan 2026 · 19:29	24 Jan 2026 · 19:10
MACROSCOPIC APPEARANCE			
Colour	Yellow	Red	Amber
Turbidity	Clear	Bloody	Opaque
Specific gravity	1.010–1.030	1.015	1.015
pH	5.0–8.0	5	6
DIPSTICK URINALYSIS			
Haemoglobin	Negative	++++	++++
Nitrites	Negative	Negative	Positive
Leucocyte esterase	Negative	+++	+++
Protein	Negative	++++	++++
Glucose	Negative	Negative	Negative
Ketones	Negative	Negative	Negative
Urobilinogen	Negative	Negative	Negative
Bilirubin	Negative	Negative	Negative
URINE SEDIMENT			
Erythrocytes	< 5/hpf	Massive	100–200/hpf
Leucocytes	< 3/hpf	10–25/hpf	100–200/hpf
Bacteria	None	Negative	++++
Origin of haematuria	—	Non-glomerular	Indeterminate

Table 1. Evolution of the urinary sediment characterized by persistent hematuria, increasing leukocyturia, and the appearance of bacteriuria with positive nitrituria.

CONCLUSION

Hemorrhagic cystitis may be a rare extrapulmonary manifestation of SARS-CoV-2, even in neonates.

COVID-19 should be considered in the differential diagnosis of macroscopic hematuria with negative urine culture to avoid unnecessary antibiotic use.

SARS-CoV-2-related urothelial injury may predispose to secondary UTIs, requiring clinical vigilance.

REFERENCES

- [1] Tiryaki S, et al. COVID-19 associated lower urinary tract symptoms in children. J Pediatr Urol. 2022;18(5)
- [2] Creta M, et al. SARS-CoV-2 infection affects the lower urinary tract and male genital system: A systematic review. J Med Virol. 2021;93(5):3133–3142 .



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Introduction

- **Overgrowth syndromes** are a broad **spectrum** of paediatric diseases mainly caused by **mutations** at different levels in **growth signalling pathways**
- Prevalence is **rare** and estimated around **less than 1 in 1'000'000**
- Clinical presentations are **variable** due to **mosaicism**
 - Example of symptoms : Excessive growth of one or more limbs or head, lipomatous/vascular malformations, dysmorphic features
 - Confounding symptoms : Common neonatal metabolic disease, macrosomia

Case report

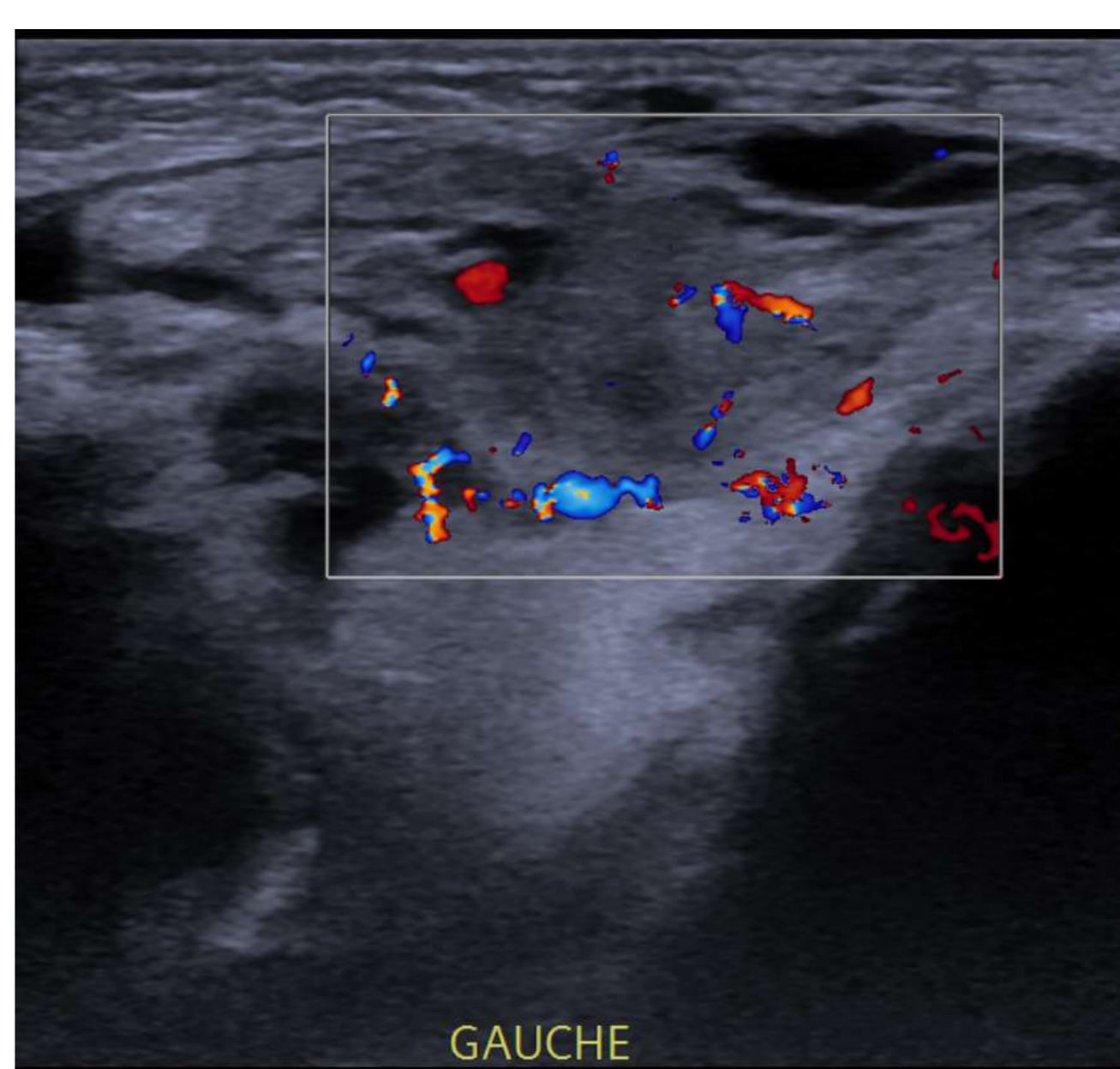
A full-term newborn boy presents **abnormalities in the upper limbs**, including **hypertrophy of the first three fingers of the right hand** and of the **first two fingers of the left hand**, **malrotation of both middle fingers**, **hypertrophy of both forearms** and **a soft lump on the left side of the neck**.



- **Pregnancy** : Insulin-dependent maternal diabetes and PCOS.
 - **Ultrasounds** : Abnormal in first semester (increased nuchal translucency) and polyhydramnios since the third semester.
 - **Choriocentesis** :
 - Karyotype and array CGH : Normal
 - Trio exome sequencing : Pathogenic heterozygous variant in the F2 gene, indicating an increased risk of thromboembolic disease.
- **Delivery** : Normal
- **Physical exam** (except the anomalies mentioned before) : Normal except a physiological heart murmur and cutis marmorata

Early management in the maternity ward

- **Malformation screening** :
 - X-Ray (Hands)
 - Ultrasounds (Abdominal, cerebral, spinal, cervical)
- **OAE test** : Pass in the right ear, fail in the left ear
- **Expert opinions** : Geneticists, metabolists, orthopaedists, cardiologists



So, what does my baby have, Doctor?

Suspected diagnosis : PIK3CA-related overgrowth syndrome (PROS)

And ... what is it?

- Activation of one (or more) of the **gain-of-function pathogenic variants** in **PIK3CA gene**
 - Mostly sporadic
 - Congenital or early onset
- **Commonly found clinical features** :
 - Overgrowth of a tissue
 - Vascular and/or lymphatic malformation
 - Cutaneous lesions (Naevus, keratosis, ...)
- **Differential diagnosis** : Proteus syndrome, Cowden syndrome, PTEN hamartoma tumour syndrome, MPPH syndrome, macrosomia with skin conditions, oncologic diseases, ...

So, how can you diagnose that?

Biopsy of the affected tissue is the best way to diagnose the disease

- **Difficulties** :
 - Where to perform the biopsy
 - Mosaicism (Disparity of the tissue and mutation distribution within the cells into the biopsied tissue)
 - Multiple assays

What is next then?

- Imaging (US, MRI)
- **Close follow-up by specialists** : Angiologists, orthopaedists, geneticists, dermatologists, developmental assessment +/- others
- **Treatment** : **No cure exists** but in certain case, **alpelisib** can be used

Key messages

- For **early diagnosis** :
 - **Prenatal assessment** and **postnatal clinical examination** must be performed thoroughly
 - **Extensive malformation screening** must be performed
- **Raising awareness** among professionals is essential to **ensure prompt management**