Severe Failure to Thrive, If It Is Not Only About Calories

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Case Report

- **12-month-old male** with severe Failure to Thrive, relevant medical history is noted below
- Caloric intake of **50kcal/kg/d** at admission with **restrictive and selective food intake**
- **Physical examination** showed a malnourished infant with mild abdominal distension but was otherwise normal

Interventions / Therapy

- **Multidisciplinary management**
- Nutrition via **nasogastric tube,** gradually increased to **95kcal/kg/d**
- **electrolyte supplementation**: sodium 11mmol/kg/d, potassium 5mmol/kg/d, chloride 16mmol/kg/d
- **NSAID** therapy administered, resulting finally in rapid catch-up in growth, as shown below

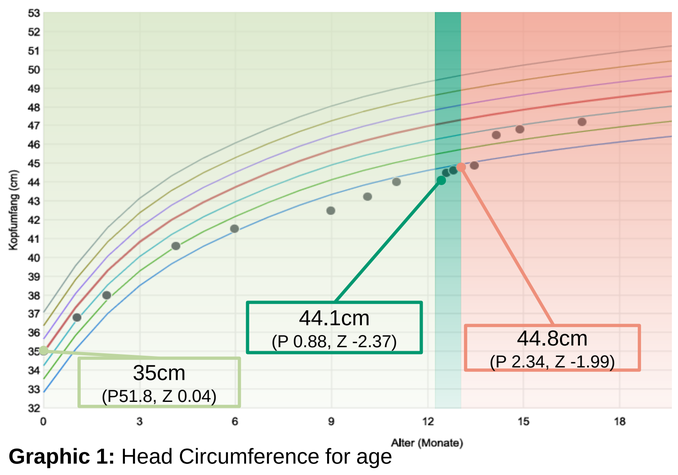
Laboratory Testing

- **Blood gas analysis** revealed a severe **hypochloremic, hypokalemic metabolic alkalosis with hyponatremia**
- **Urine analysis** indicated **renal salt loss**
- Liver function tests, inflammatory markers, thyroid and parathyroid levels, vitamins, micronutrients, complete blood count, gastrointestinal antibodies, hormone levels, sweat test and metabolic analyses showed no major abnormalities
- diuretic use and gastrointestinal losses were ruled out

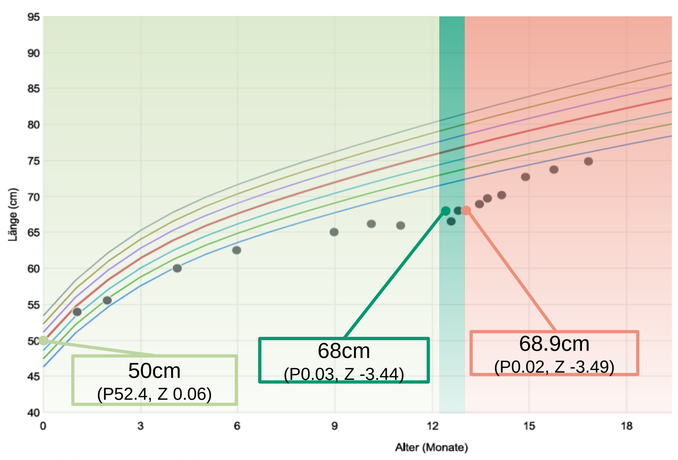
pH	7.66↑↑
pCO2	34
Bicarbonate	38.6 ↑
Base excess	15.6 ↑
pO2	58
p50	24
Sodium	130 ↓
Potassium	2.1↓↓
Chloride	85 ↓
Calcium total	2.76 ↑
Calcium alb.-corr.:	2.73
Inorg. Phosphate:	1.41
Magnesium:	1.08 ↑
Glucose	6.2

Table 1: Initial venous BGA

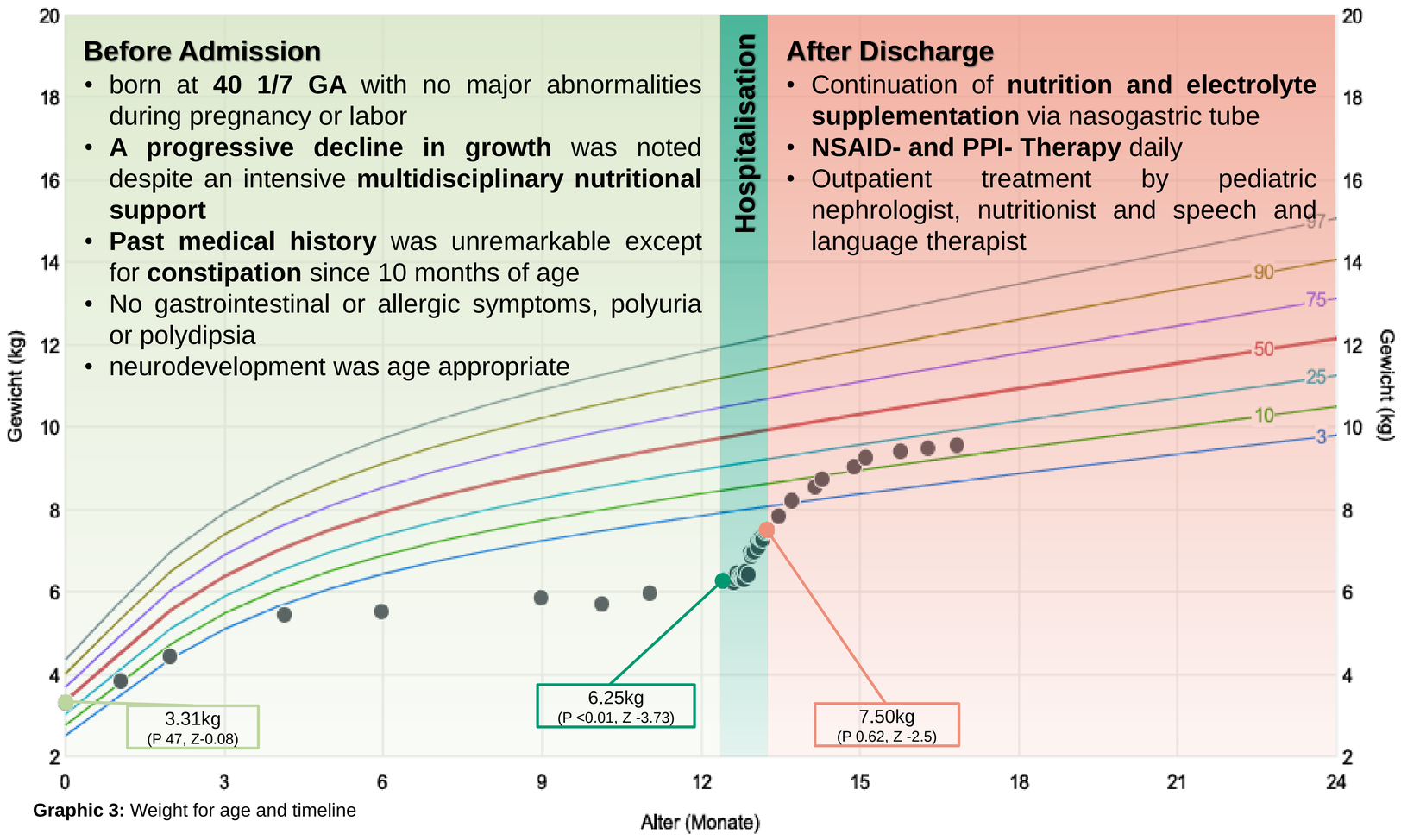
Genetic testing revealed compound heterozygous pathogenic variants in the CLCNKB gene, confirming the diagnosis of Bartter syndrome type 3



Graphic 1: Head Circumference for age



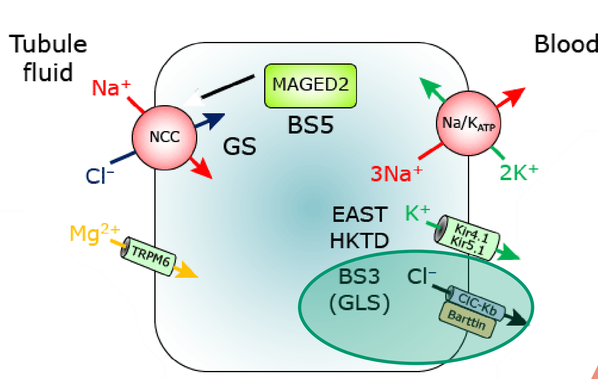
Graphic 2: Length for age



Graphic 3: Weight for age and timeline

Discussion

- Failure to thrive is common and a **symptom rather than a diagnosis**
- **Possible underlying causes** are diverse and may include inadequate caloric intake, malabsorption, metabolic disorders, increased losses or excessive energy expenditure [1]
- **In Bartter Syndrome type 3** the **pathogen CLCNKB gene** encodes the **ClC-Kb channel** in the distal convoluted tubule, impairs the renal chloride reabsorption and leads to a **renal salt loss**, a **hypokalemic metabolic alkalosis**, normal to low blood pressure, normal magnesium levels, hyper- to normocalciuria and absence of nephrocalcinosis [2]
- It typically presents in late infancy or early childhood with **failure to thrive, polyuria and polydipsia, dehydration, salt craving, constipation, hypotonia and recurrent vomiting** [4]



Graphic 4: Schematic diagram of distal convoluted tubule cell and ClC-Kb Chanel [3]

Conclusion

- This case highlights that renal salt-wasting disorders are a **rare but important cause** of failure to thrive, **even without classic presentation**
- Early recognition is crucial, as **targeted therapy can lead to rapid and substantial catch-up growth**, as shown above

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COLLAGENOUS GASTRITIS, A RARE DISEASE IN REFRACTORY IRON DEFICIENCY

Clinical Case Presentation | Author and Presenter: Rosi-Wumba Lusua; Co-authors: Prof. Andreas Nydegger, Prof. Christine Sempoux, Giovanna Alfano | Institution: Département femme-mère-enfant - DFME CHUV

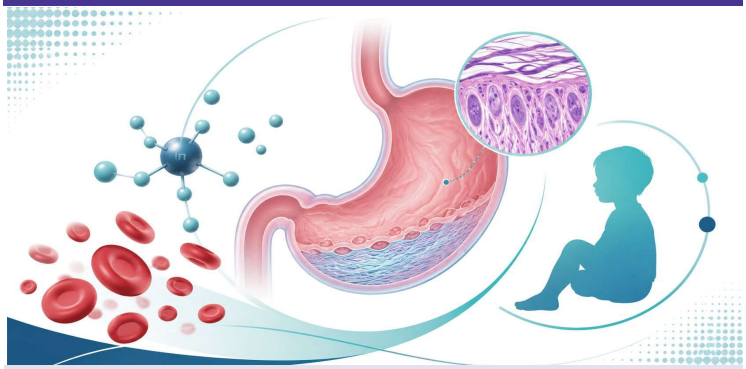
BACKGROUND

Collagenous gastritis (CG): extremely rare histological entity — subepithelial collagen bands >10 µm in gastric mucosa.
Fewer than 300 pediatric cases ever reported in Europe. Pathophysiology poorly understood; Symptoms may be mild or absent, contributing to delayed diagnosis. No standard therapy established; treatment is individualized. Options include diet modification, topical budesonide, sucralfate, and mesalazine

Two Clinical Forms

- **Pediatric form:** predominantly girls, iron-deficiency anemia, often without digestive symptoms, isolated disease (not associated with colitis)
- **Adult form:** associated with collagenous colitis and autoimmune diseases; more varied clinical presentation

CASE REPORT



- **10-year-old girl initially hospitalized in oct. 2024 for severe anemia,** Subsequently followed by Hematology for: Pancytopenia Severe hypochromic microcytic anemia
- Referred to Gastroenterology for further investigations for **persistent iron deficiency refractory** to oral supplementation.
- Mild intermittent crampy abdominal pain;
- **No overt gastrointestinal bleeding** (no hematemesis, melena, or hematochezia)
- No diarrhea; or weight loss; No other bleeding symptoms
- **Clinical examination unremarkable**

Key Laboratory Values

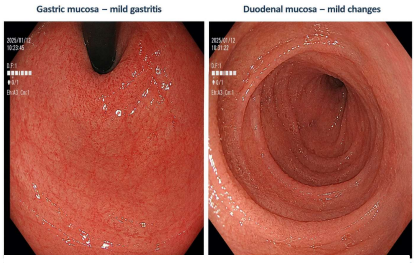
- **Hemoglobin: 45 g/L** (ref >120 g/L) **Platelets (min): 140 G/L (ref 150-400 G/L)**
- Plasma iron: 4.0 µmol/L (ref 9-30) **Ferritin: 9.0 µg/L** (ref 12-150) TSAT: 6% (ref 20-45%)
- **IgA deficiency limiting interpretation of celiac serology**
- Calprotectin: negative

Exclusions

- Hemoglobinopathies (thalassemia, zinc protoporphyrin): negative
- Viral serologies (CMV, HSV, EBV): negative; Parvovirus IgG+/IgM−
- Hemolysis (haptoglobin), renal/hepatic function: normal

DIAGNOSTICS: ENDOSCOPY & HISTOLOGY

Endoscopy — January 2026

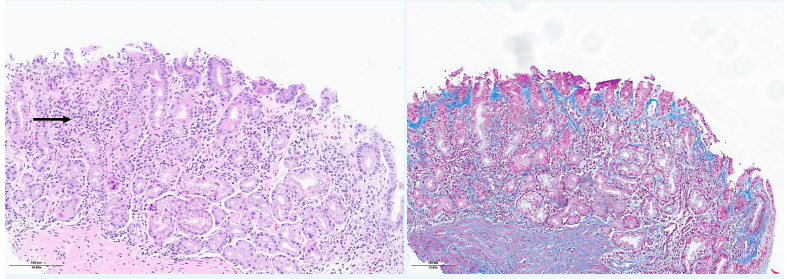


Stomach: diffuse nodularity + erythema/hyperemia, without hemorrhage
Duodenum: focal atrophy, cobblestone appearance (bulb + D2)
Esophagus: normal mucosa

Atypical findings initially raised suspicion for celiac disease and H. pylori.

Histology of Gastric Biopsies

H&E (hematoxylin-eosin stain) *stain (x15.84)* *Masson trichrome (x13.62) - collagen in blue*



- Gastric mucosa with mild diffuse chronic inflammation and surface damage
- Lymphoplasmacytic infiltration with reactive lymphoid follicles
- **Thick subepithelial collagen bands** — hallmark finding
- Superficial erosions; **H. pylori: NOT DETECTED**
- Duodenal biopsies was negative for celiac disease

→ Consistent with Collagenous Gastritis

MANAGEMENT & FOLLOW-UP

- She is now on Iron supplementation (partial response); PPIs initiated post-endoscopy based on macroscopic findings.
- **gluten-free diet**
- Follow-up gastroscopy in July to evaluate gluten-free diet response and gastric mucosal recovery.
- multidisciplinary approach (hematology, gastroenterology, pathology) required.

CONCLUSIONS AND KEY POINTS

This case highlights the importance of diagnosing rare conditions like collagenous gastritis in the context of refractory iron deficiency.

Endoscopy combined with histologic assessment remains essential for accurate diagnosis.

Further studies are needed to better understand this pathology and develop more effective and validated treatments.

EARLY RECOGNITION. ACCURATE DIAGNOSIS. INDIVIDUALIZED CARE. — TO IMPROVE OUTCOMES FOR CHILDREN WITH RARE DISEASES.

When pediatric acute abdomen needs a deeper glance at renal function

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Background

Acute abdominal pain in children is frequent and encompasses a wide spectrum of age-dependent etiologies, ranging from benign self-limiting conditions to severe causes[1]. Identifying severely affected patients remains a clinical challenge. In children with nephrotic syndrome spontaneous bacterial peritonitis (SBP) due to ascites, hypoproteinemia and impaired immune function represents a potentially life-threatening complication. Nephrotic syndrome is the most common first manifestation of pediatric glomerulopathy[2, 3, 4].

Case Study

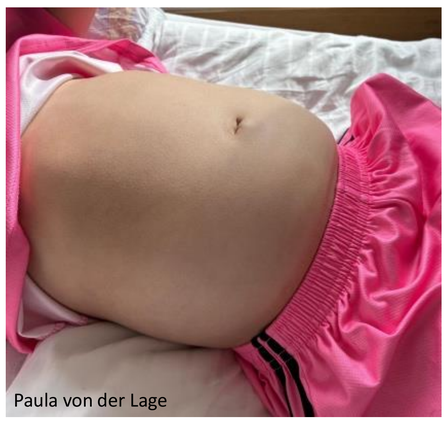

Figure 1

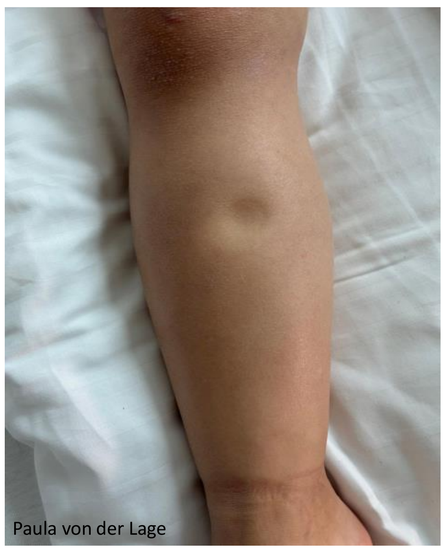

Figure 2

A 6-year-old girl presented to the pediatric emergency department with severe abdominal pain and bilateral leg edema (Figure 2). Clinical examination showed markedly reduced general condition with a distended abdomen (Figure 1), bilateral pretibial and periorbital edema, fever (40°C), tachycardia (150/min), oxygen saturation of 97% and arterial hypertension (131/90 mmHg). Laboratory findings revealed initially a C-reactive protein of 98 mg/L and a procalcitonin of 74 ng/mL. Abdominal ultrasound demonstrated moderate ascites, while computed tomography showed no other causes of acute abdominal pain. Urine albumin-to-creatinine and protein-to-creatinine ratio peaked at 1277 mg/mmol and 2766 mg/mmol, respectively.

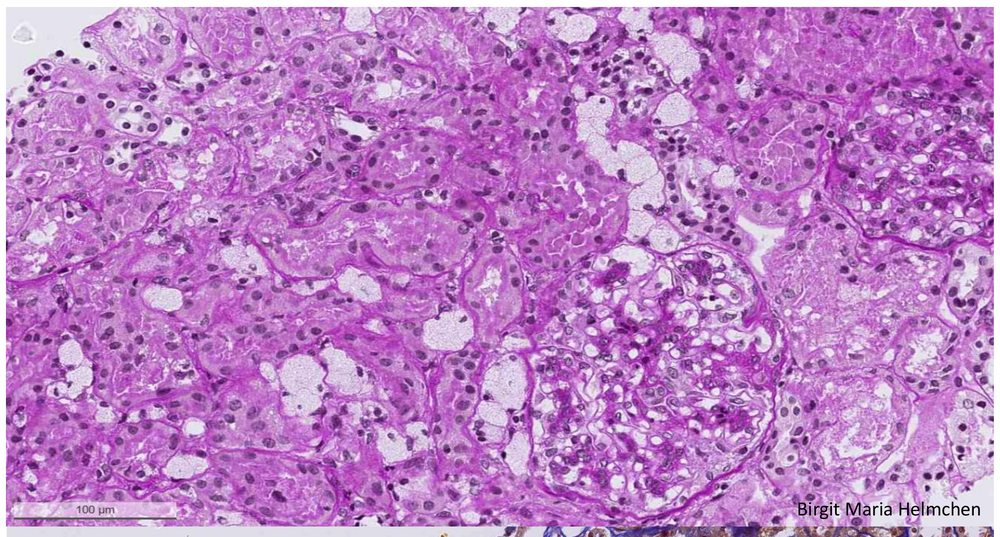

Figure 3

Alltogether, symptoms were explained by sepsis due to SBP as a complication of a newly diagnosed nephrotic syndrome. Urine sample collection was challenging and took some time. Focused anamnesis and reassessment of characteristic symptoms, some of them present for a few weeks before admission, supported the diagnosis. A favorable clinical course was observed on antibiotic and corticosteroid therapy. Persistent proteinuria despite corticosteroid therapy, led to the diagnosis of steroid-resistant nephrotic syndrome[3]. Subsequent renal biopsy resulted in focal segmental glomerulosclerosis (FSGS) (Figure 3). A treatment with tacrolimus was initiated. The family also attended a genetic counseling consultation[3, 4, 5].

Summary

Edema, a hallmark of nephrotic syndrome, was a key clinical finding suggesting the medical etiology for acute abdominal pain in this case[3]. Urinalysis, in combination with fever and ascites, ultimately led to the correct diagnosis. FSGS represents the second most common underlying histopathology of nephrotic syndrome and is associated with a high risk of progression to chronic kidney disease. FSGS with persistent proteinuria requires immunosuppressive therapy in most cases[1, 2, 3, 5].

Conclusion

In children presenting with acute abdominal pain and edema, nephrotic syndrome should be considered as an underlying condition[1, 4]. As a complication of nephrotic syndrome, SBP represents an important etiology of pediatric acute abdomen[4, 6]. Early recognition allows prompt initiation of antibiotic and corticosteroid therapy and may prevent further deterioration or unnecessary surgical interventions. This case underlines the importance of thorough assessment for accurate diagnosis.

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Enteric duplication cysts : a rare diagnosis in the newborn

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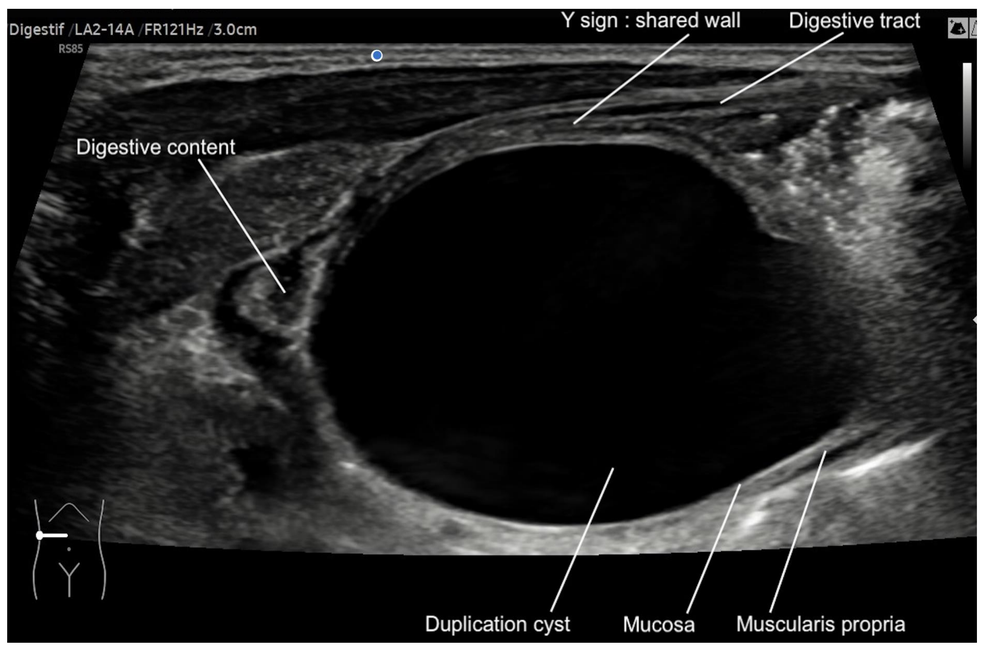
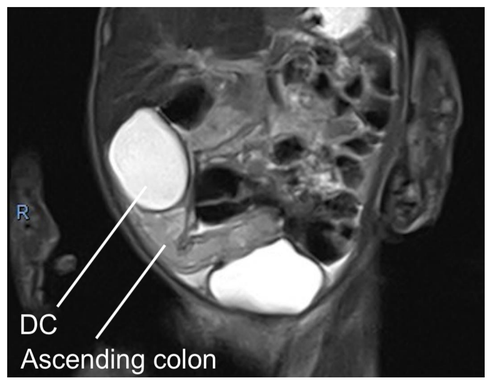
Introduction

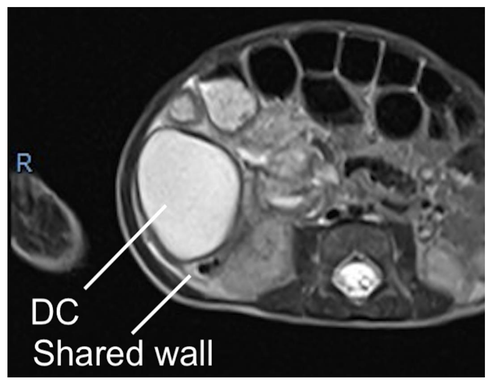
- Enteric duplication cyst (EDC) is a **rare congenital malformation**.
- It may occur throughout the digestive tract and is most commonly located in the ileum and oesophagus, located on the mesenteric aspect of the intestine.
- The **most frequent form is cystic** (75%), with a smooth muscle wall and common blood supply, and can contain heterotopic gastric mucosa.
- Many are discovered incidentally or through prenatal diagnosis (37-70% of cases) and **can be asymptomatic**. Some are associated with vertebral anomalies.
- **Clinical presentation is highly variable** depending on the complications : abdominal pain, obstructive presentation, bleeding, intussusception, inflammation, perforation, or malignant transformation.

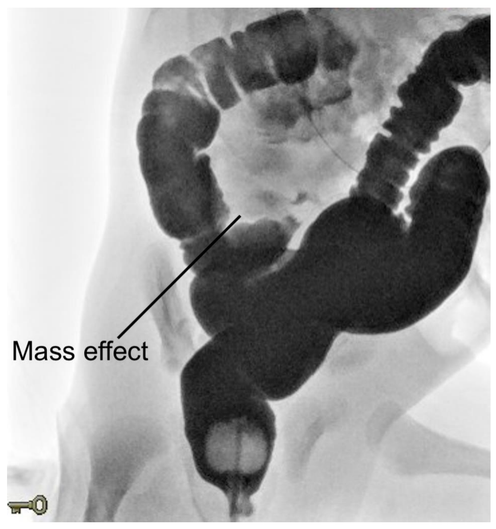
Case Report

- **12-day-old** female infant, born at term, known for intrauterine growth restriction without antenatal abnormalities **presented with bilious vomiting and failure to regain birth weight.**
- On clinical examination, she was alert, comfortable and active, without fever. Abdominal palpation revealed a mass in the right flank.
- Ultrasound and MRI were performed and revealed a compressive duplication cyst of the ascending colon.
- She was transferred to university hospital for further management. Primary decision was to postpone surgery because of a subocclusive presentation.
- Due to recurrence of bilious vomiting and obstructive symptomatology, surgical intervention was performed at 28 days of life : **marsupialisation of the cyst without enteric resection (for ileocecal valve conservation).**
- Post-operative follow-up was successful with gain of weight after refeeding.









Radiological findings

- Gut signature sign is an **inner hyperechoic mucosa, outer hypoechoic smooth muscle layer**. EDC shares the wall of **adjacent loop.**
- EDC of ileal or colonic origin causes **upstream intestinal dilatation**.
- **Differential diagnosis**: torsioned ovarian cyst, mesenteric cyst, Meckel's diverticulum (in this case, ovaries not visualized).
- Abdominal MRI further characterized the lesion as a duplication cyst of the ascending colon with mass effect.

Conclusion

- This case highlights the importance of considering an enteric duplication cyst in newborns presenting with bilious vomiting associated with a palpable abdominal mass.
- The method of choice to diagnose EDC is ultrasonography.
- Surgical treatment with early complete excision is preferred to avoid future complications.

References

RECOGNIZING JUVENILE MYOCLONIC EPILEPSY IN ADOLESCENTS: EPIDEMIOLOGY, CLINICAL FEATURES, DIAGNOSTIC PITFALLS AND THE ESSENTIAL ROLE OF TARGETED HYSTORY-TAKING

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INTRODUCTION & BACKGROUND

WHAT IS IT JUVENILE MYOCLONIC EPILEPSY?

A hereditary, idiopathic generalized epilepsy

Bilateral myoclonic jerks, mainly on awakening, that are usually isolated for months to years.

Generalized tonic-clonic seizures (**GTCS**) appear +/- 3 years later, less frequently **absence seizure**

COMMON TRIGGERS

Sleep deprivation

alcohol

stress

EPIDEMIOLOGY

JME accounts for **5-10%** of all epilepsies

Typically begins **between 12 and 18 years of age**

Prevalence of 10-20 cases / 100.000

Female to male ratio 1 to 1.8:1

Diagnostic delay ranges from 3 to more than 10 years

JME is **frequently underdiagnosed** because subtle myoclonic jerks are overlooked, misinterpreted or not actively investigated, leading to diagnostic delay.

The aim of this poster is to highlight the importance of **targeted history-taking** in adolescents presenting with GTCS and to illustrate common causes of diagnostic delay in JME through two clinical cases.

CASES & TIMELINES

CASE 1 — 17 YEAR OLD MALE

Time to diagnosis: 8 months

1. Reported history of sudden upper limb jerks with objects dropping and transient knee buckling
2. Misdiagnosis of stress related fasciculations
3. Facial trauma after unexplained fall
4. Normal cerebral IRM
5. First observed GTCS at awakening time
6. Targeted history taking and targeted EEF
7. Diagnosis of JME
8. Levetiracetam and adequate seizure control

CASE 2 — 17 YEAR OLD MALE

Time to diagnosis: 3 years

1. Morning myoclonic jerks, dropping objects
2. GTCS after alcohol consumption → Diagnosed with provoked GTCS
3. 1 Normal EEGs remotely from the ictal event
4. Persistent myoclonic symptoms
5. Normal cerebral IRM
6. Second GTCS with alcohol intake
7. Targeted history taking and targeted EEG
8. Diagnosis of JME

EEG FINDINGS

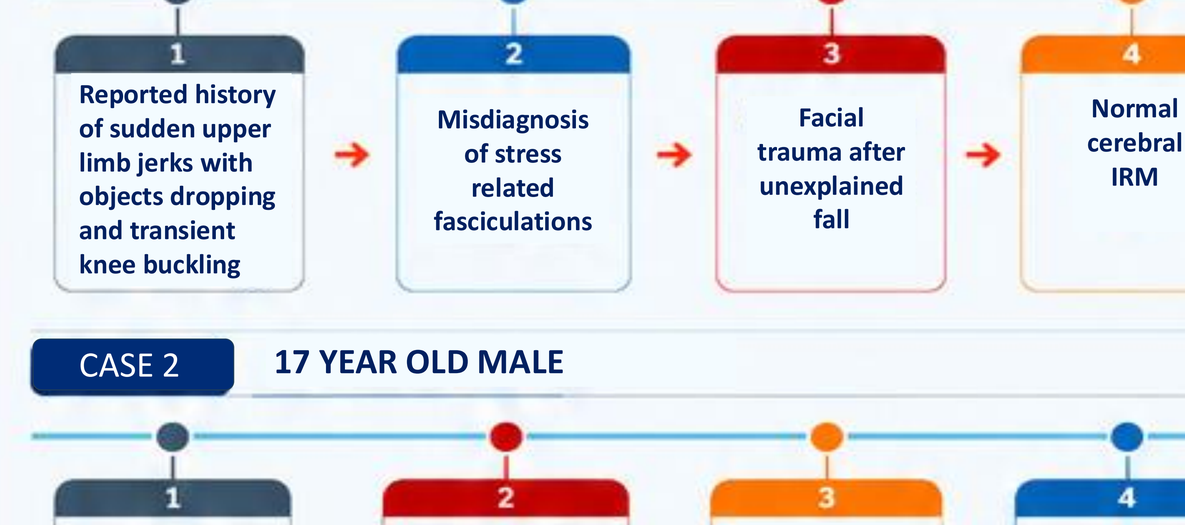
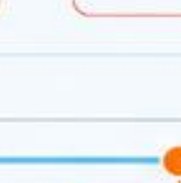


Illustration reproduced with permission from Dr. Bassi : spike-waves discharges in sleep-wakefulness transition phases

TAKE HOME MESSAGES

Subtle myoclonic symptoms are often unrecognized or minimized (stress, fatigue, clumsiness) → we need to **actively identify them**

The gold-standard tool for diagnosis is EEG.
Routine awake EEG has limited sensitivity (~73%).
Sleep deprivation EEG increases the sensitivity up to 100%



Brain MRI is usually normal in JME and is **not routinely required**

Early recognition prevents delays (3.5-14.5 years on average) and allows appropriate long-term treatment

KEY QUESTIONS TO ASK

Do you drop objects unexpectedly?

Are your symptoms worse when you are sleep-deprived or very tired?

Do you have sudden jerks of your arms or shoulders in the morning?

Do your knees buckle or do you lose tone briefly on awakening?

A Complex Bleeding

Hemoptysis and Apparent Epistaxis as Manifestation of a Congenital Arterial Fistula

Mattesini B E [1], Vandelli A C [2], Despont F [3], Anguissola G [3], Rizzati F [3], Rybak A [2], Rouge Elton P [2]

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Case : A 10-month-old infant with no significant medical history was admitted after experiencing four episodes of hemoptysis, two of which being accompanied by nasal bleeding, resolving spontaneously within a few minutes, in the context of a dry cough and nasal congestion for the previous 10 days.

At admission in our pediatric emergency department, he was hemodynamically stable. He presented dried blood in the nasal cavity, without other significant findings. Blood tests showed moderate anemia (9,9 g/dL) with no signs of coagulopathy and the chest radiography was normal. In the absence of identifiable nasal lesion on ENT fiberoptic examination, a lower respiratory tract origin was suspected and the patient was hospitalized for observation.

He subsequently experienced two other episodes of bleeding, with a 2 g/dL drop in hemoglobin, requiring nebulized tranexamic acid administration. Urgent bronchoscopy revealed a lesion on the bronchial mucosa of the right lower bronchus with active bleeding. Hemostasis was achieved by packing and the patient received a transfusion of erythrocyte concentrate. The patient was admitted in the pediatric intensive care unit for invasive ventilation.

An injected thoracic CT scan performed 24 hours after admission showed areas of alveolar hemorrhage in the right lower and middle lobes (Fig.1) and suggested the presence of a collateral artery originating from the right subclavian artery, identified as the bleeding source.

Angiography revealed the presence of an arterio-arterial fistula between the right bronchial artery and the postero-basal subsegmental pulmonary artery of the right lower lobe (Fig.2A). The infant successfully underwent embolization (Fig.2B; Fig.3).

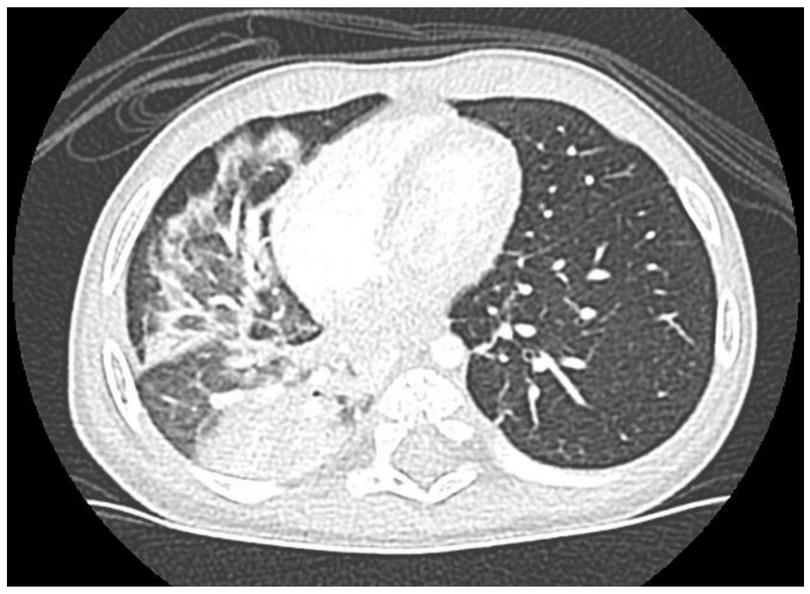

Fig.1: Injected thoracic CT-scan: areas of alveolar hemorrhage in the right lower and middle lobes, appearing as ground-glass opacities.

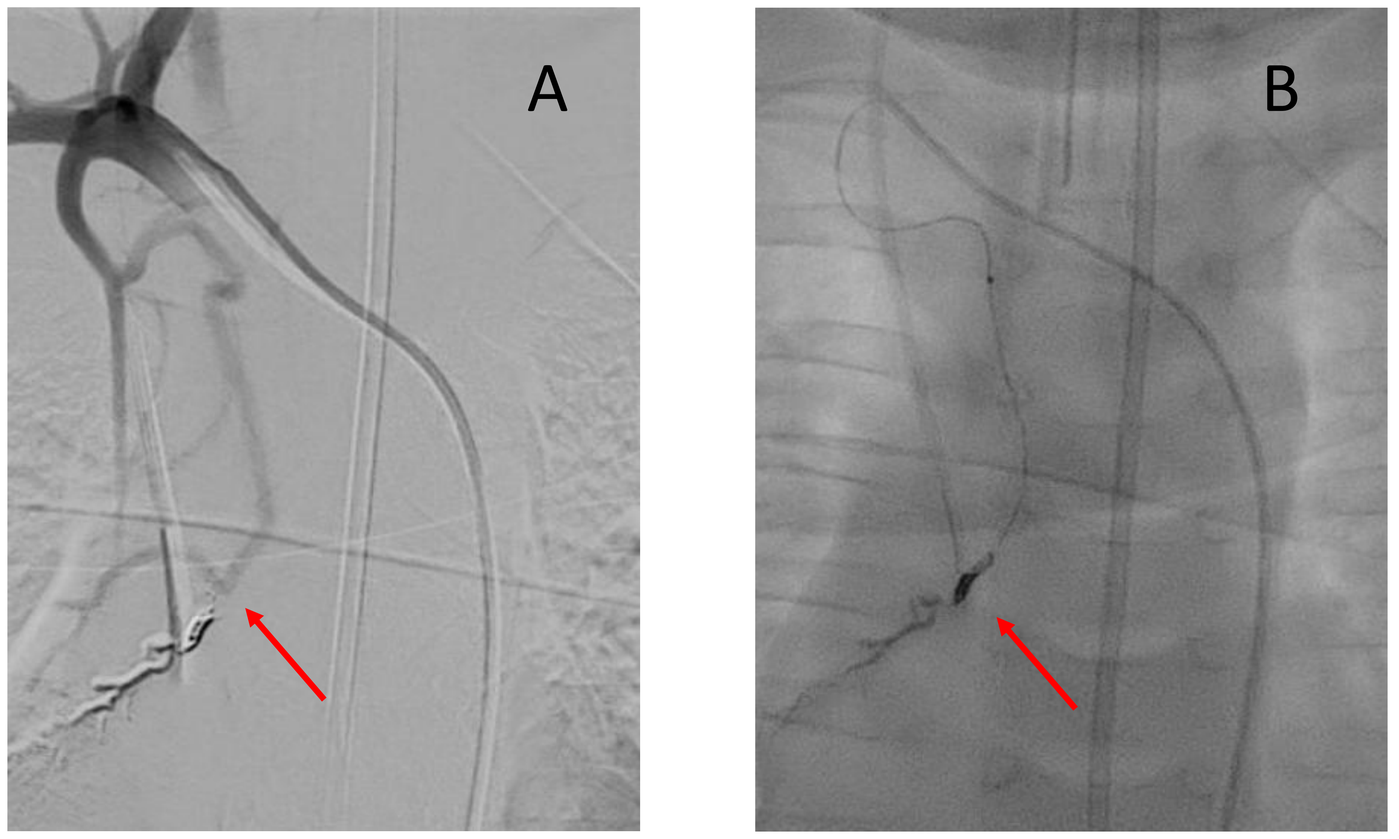

Fig.2: Angiography:
A) Microcatheterisation of the right bronchial artery demonstrating an arterio-arterial fistula between this bronchial artery and a subsegmental branch of the segmental artery of the posterobasal segment of the right lower lobe.
B) Microcatheterisation of the superior branch of this arterio-arterial fistula and embolization with a coil.

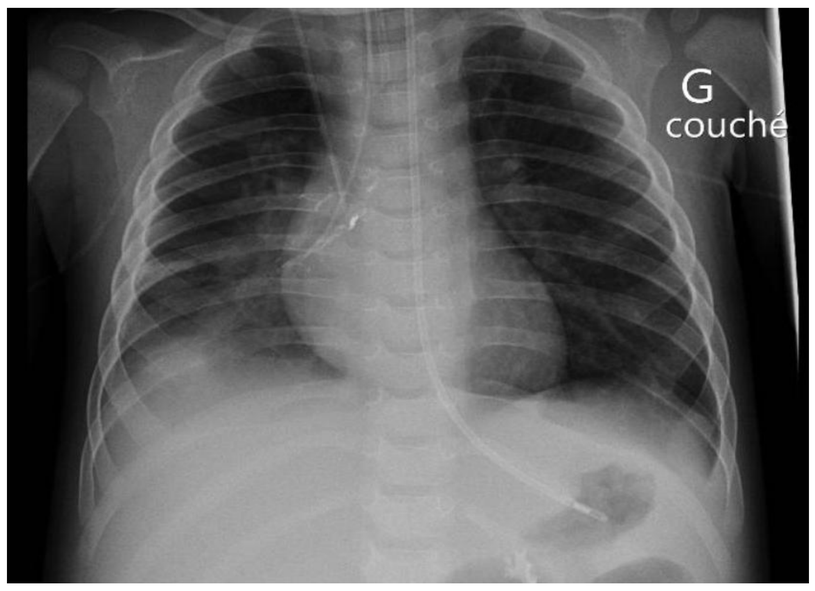

Fig.3: Chest X-ray post embolization: the coil in the proximal branch of the fistula.

Respiratory tract bleedings in infants are mainly represented by epistaxis and in most cases are related to upper respiratory infections and traumatic nasal irrigation with saline solution done by parents

However, nasal bleeding, especially if accompanied by hemoptysis, may originate from the lower respiratory tract

Take Home Message → Do Not Underestimate Airway Bleedings!

Congenital vascular malformations of the airways may manifest through recurrent hemoptysis and nasal bleedings

Early diagnosis and timely intervention is important to prevent life-threatening complications.