HIDDEN FOR YEARS – SEEN IN SECONDS

From Chronic Pain to Surgical Emergency

Département de pédiatrie

Toumi L,
Calinescu AM*,
Spigariol F

*Geneva University Hospitals,
Pediatric Surgery Unit

Introduction

- Abdominal pain accounts for ≈ **12 %** of pediatric emergency consultations
- 90 % **Non-Surgical** cases
- **Functional and chronic pain** are highly prevalent
- **Internal Hernia and bowel ischemia** :
  - **Rare** in children
  - Clinical and laboratory findings are often **non-specific**
  - **Challenging** to diagnose

Case Presentation

Clinical Course

Functional Abdominal Pain → **Surgical Abdomen**

10-year-old

- **Recurrent abdominal pain**
- **No** Red Flags
- Exclusion of celiac disease and inflammatory bowel disease

12-year-old

- **ED consultation for intense pain episode**
- Normal ultrasound
- Spontaneous remission

14-year-old

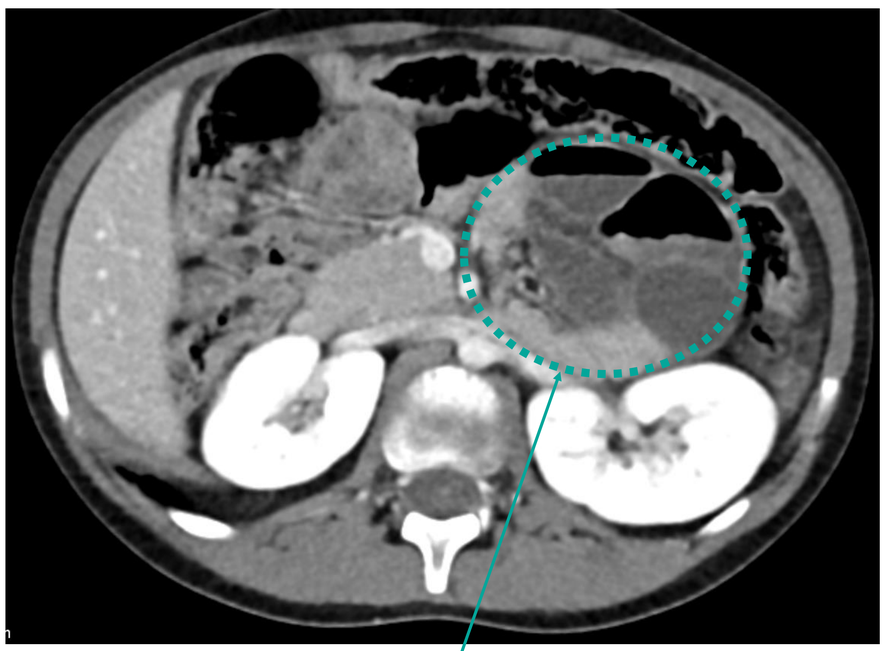
- **Unusual, severe abdominal pain**
- Unwell appearance and hypertensive
- Diffuse abdominal tenderness, bowel sounds preserved
- No relief despite opioid treatment

Diagnostic Work-Up & Outcome

Nonspecific ultrasound

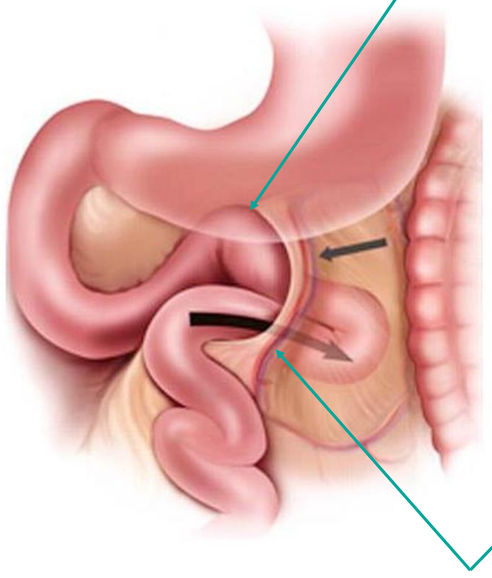
Normal laboratory including lactate

Urgent abdominal CT-Scan: Incarcerated left paraduodenal Hernia



Small bowel in the left para-duodenal hernia sac

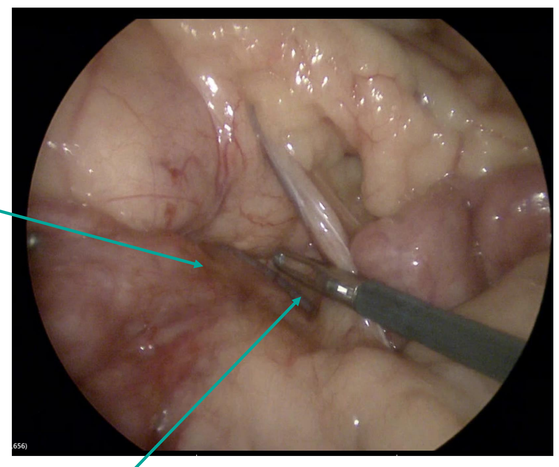
Anatomic landmarks



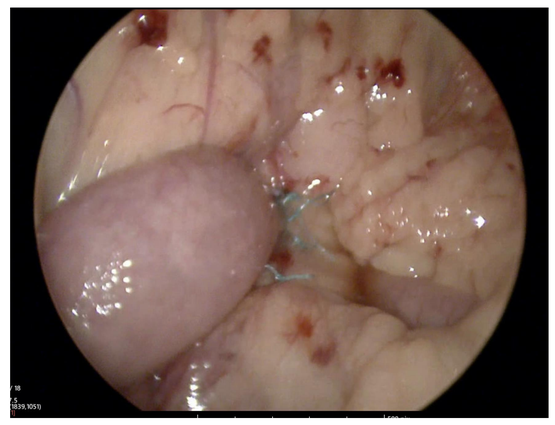
Duodeno-jejunal junction

Inferior mesenteric vein

Emergency surgery



Closure Bowel preserved



Discussion & Literature

Contact: leila.Toumi@rhne.ch

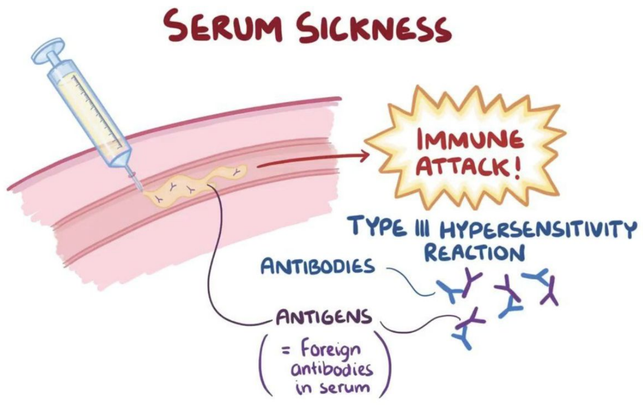
- Functional diagnosis must remain open to **re-evaluation** in evolving symptoms
- Normal labs and non-specific ultrasound **do not rule out** surgical pathology
- **Imagery escalation** is required in clinically severe presentations
- **CT-Scan is key** to diagnosing bowel ischemia and internal herniation
- **Time-critical recognition** of internal hernia is essential to prevent bowel loss

Serum sickness-like reaction: a pediatric case report

Swiss Paediatrics Congress | 11-12 June 2026

Authors : Maresca A., Luna De Haro C., Zellali K.
Department of Pediatrics, Hôpital Intercantonal de la Broye, site de Payerne, Switzerland

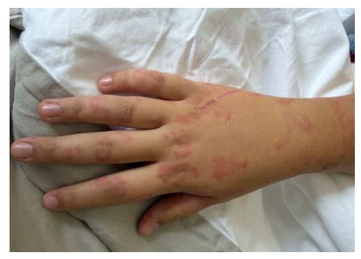
Alessandro Maresca | Hôpital Intercantonal de la Broye, site de Payerne



Serum sickness–like reaction (SSLR) is a rare immune-mediated hypersensitivity reaction characterized by urticarial or target-like skin lesions, arthralgia and systemic symptoms. Post-vaccination SSLR is extremely rare in pediatric patients

CASE PRESENTATION

- Previously healthy 13-year-old presenting with fever and an acute, intensely pruritic rash (figures 1-5)
- Rapid evolution to target-like urticarial lesions with facial and palmoplantar edema (figures 6-10)
- Abdominal pain and bilateral arthralgia requiring hospitalization
- Symptom onset 15 days after first MenB (Bexsero®) vaccination



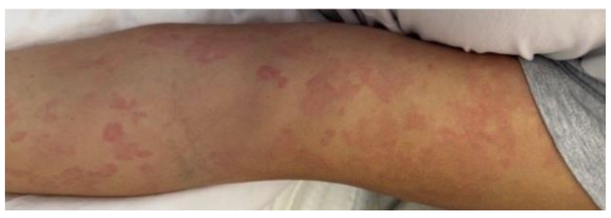
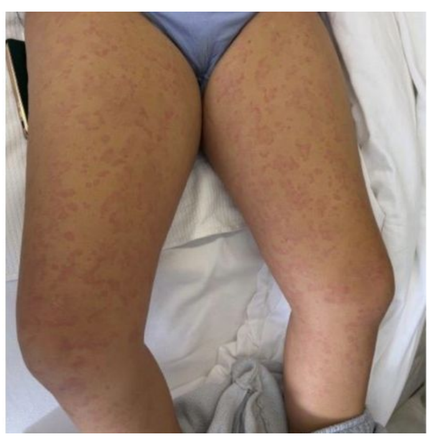
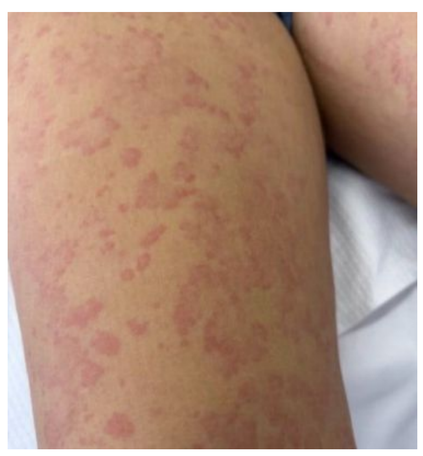
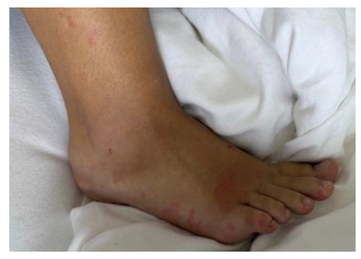


Figure 1: urticarial plaques of the arm during the acute phase



Figures 2 & 3: symmetrical urticarial plaques of the lower limbs during the acute phase

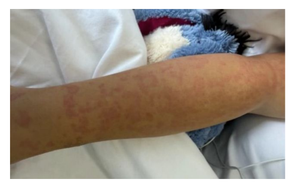


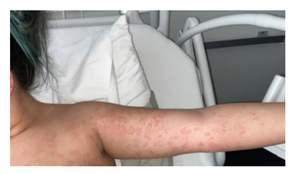


Figures 4 & 5: urticarial plaques of the hand and the foot during the acute phase

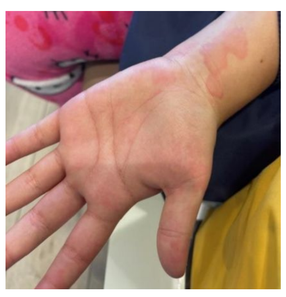
INVESTIGATIONS

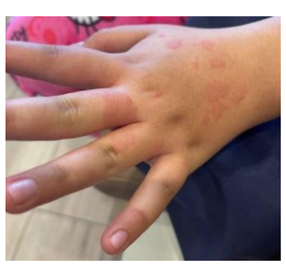
- Leukocytosis and elevated C-reactive protein - Complement was normal
- Mild persistent proteinuria. No hepatic involvement. Hemodynamically stable throughout hospitalization





Figures 6 & 7: urticarial plaques of the arms during the subacute phase

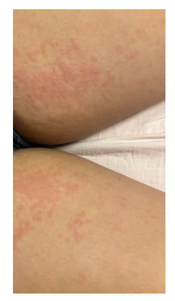




Figures 8-10: urticarial plaques of the hands during the subacute phase

MANAGEMENT & OUTCOME

- Diagnosis of serum sickness–like reaction established
- Treatment with systemic corticosteroids (prednisolone 1 mg/kg/day for 5 days) and antihistamines
- Rapid clinical improvement; brief recurrence of urticarial lesions (figures 11 & 12)
- Complete recovery without sequelae



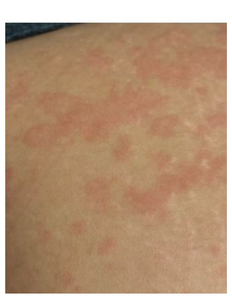


Figure 11: symmetrical urticarial plaques of the lower limbs during the remission phase

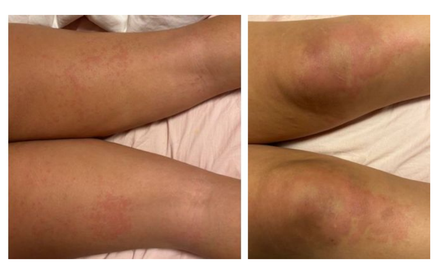


Figure 12: symmetrical urticarial plaques of the lower limbs during the remission phase

CONCLUSION

SSLR should be considered in children presenting with urticarial eruptions and systemic symptoms. Vaccination may represent a rare triggering factor. Pharmacovigilance reporting to Swissmedic is ongoing.

References:
C.C., E.K., P.A., I.E., V.K., T.P., D.S., M.-E. T., K.V., D.Z., V.M., P.G., P.E., L.L. and D.A.
Serum Sickness-Like Reaction: A Narrative Review of Epidemiology, Immunopathogenesis, Diagnostic Challenges and Therapeutic Approaches - PMC
Clin Pract. 2025 Sep 26; 15(10): 178.

Avenue de la Colline 3
1520 Payerne

Rare case of a seronegative ocular Myasthenia in a 3-year-old boy.

Authors: [1] Laxy E., [1] Meier S., [1,2] Schedel A., [1] Ober H., [1] Broser P. J.

[1] Department of Pediatric Neurology; Ostschweizer Kinderspital St. Gallen; Switzerland
[2] Department of Neurology; HOCH; St. Gallen; Switzerland

Introduction

Myasthenia gravis is an autoimmune disorder of neuromuscular transmission, caused by antibodies that impair acetylcholine receptor function at the postsynaptic membrane. Pediatric myasthenia gravis is a rare condition in children, and isolated ocular manifestations in young children have been described only in a few cases in the literature. Overall, isolated ocular manifestations are more common in children than in adults. (1)

Clinical features include fluctuating ptosis, ophthalmoplegia, and diplopia and may remain purely ocular, especially in prepubertal patients.

Antibodies can be found, but there are also seronegative cases of myasthenia, which can result in a delayed diagnosis. Ice pack tests and fatigability after sustained upgaze can serve as quick bedside tests to support the diagnosis. Electrophysiological testing (NCS/EMG) is highly sensitive for detecting myasthenic disorders, abnormal findings may be associated with later generalization in ocular myasthenia, although this association has not been shown to be statistically significant. (3)

We present the case of ocular myasthenia gravis in a 3-year-old boy.

Case Report

We report a 3-year-old boy who initially presented in June 2025 with fluctuating left-sided ptosis, worsening in the evening, and suspected diplopia. Shortly before symptom onset, he had a generalized exanthem, as reported by his mother; however, a preceding viral infection remained uncertain. Initial differential diagnoses included congenital myasthenic syndrome and Miller–Fisher overlap syndrome. Over time, the ptosis became persistent and predominantly left-sided. He also developed impaired ocular motility, mainly on the left, resulting in a compensatory head posture (laterocaput). No additional neuromuscular symptoms were observed. According to his symptoms ocular myasthenia gravis was suspected. In November 2025, symptoms progressed, with the development of astigmatism, likely secondary to persistent ptosis.

The patient is from a Ghanaian family with five children, including a healthy twin sister. There is no known family history of neurological disease.

Diagnostic workup showed age-appropriate findings on brain MRI. Comprehensive ophthalmological evaluation revealed no structural ocular pathology. Cerebrospinal fluid analysis was unremarkable, including negative Borrelia serology. The ice pack test was not performed.

No myasthenia-associated antibodies were detected (AChR, MuSK, titin, LRP4, ganglioside antibodies; VGCC P/Q-type and N-type), and repeat testing during follow-up remained negative. Extended immunological testing using cell-based assays (December 2025) was also unremarkable.

Neurophysiological studies initially suggested a generalized inflammatory process. Nerve conduction studies were age-appropriate; however, F-waves were absent in the median and fibular nerves. A pathological A-wave was detected in the tibial nerve. Repetitive nerve stimulation demonstrated a decremental response in the right abductor hallucis and left orbicularis oculi muscles, indicating neuromuscular junction dysfunction. On follow-up (October 2025), no decrement was detectable, suggesting improvement.

Duo-exome sequencing (patient and mother), including analysis for mitochondrial disorders and congenital myasthenic syndromes, revealed no pathogenic variants.

Therapeutic trials with intravenous immunoglobulins (IVIG; Privigen) and pyridostigmine (Mestinon) did not result in a clinical improvement. Corticosteroid therapy (Prednisolone) initiated in October 2025 resulted in the first clinical improvement after lack of response to IVIG and pyridostigmine. In april 2026, ocular motility had normalized, and corticosteroid therapy was discontinued. Unfortunately, the symptoms recurred shortly afterwards at follow up in May 2026.

Before initiation of therapy:

July 2025
First presentation
IVIG

August 2025

Pyridostigmine

November 2025

Prednisolone

After initiation of therapy:

December 2025

Prednisolone

April 2026

Prednisolone

May 2026

Stopping: Prednisolone

Discussion

While literature on generalized myasthenia gravis (MG) is extensive, pediatric ocular MG remains less characterized with only a limited number of cases described. Our case shares clinical parallels with existing reports, such as unilateral ptosis as an initial presentation and the subsequent development of ophthalmoplegia and diplopia. Similar to the referenced cases, the association with prior viral illness is frequently observed; however, our patient's presentation with an exanthem was not clearly associated with a viral infection.

Diagnostic considerations for pediatric ocular MG are challenging. The ice pack test is a valuable diagnostic tool, though its feasibility in toddlers may be limited by patient compliance. Furthermore, although chest MRI is the gold standard for thymoma screening, exposure of anaesthesia in young children remains a concern. In our case, chest X-ray and MRI initially showed normal findings. To ensure an ongoing screening for thymoma another chest MRI is planned.

Serological testing is essential; however, the high rate of seronegativity in pediatric patients underscores the need for comprehensive screening, including MuSK and LRP-4 antibodies, which are often omitted in initial panels. In cases of persistent seronegativity, differential diagnosis must include Congenital Myasthenic Syndromes (CMS). Unlike the autoimmune MG form, CMS is identified through genetic sequencing. In our patient, exome sequencing was negative, effectively ruling out CMS.

Regarding treatment, pyridostigmine and IVIG are effective initial interventions, though clinical response can be delayed - as seen in reported cases where improvement took up to four months. (2) Finally, a recent systematic review highlights that up to 39% of adult ocular MG patients progress to generalized MG, with AChR-positivity as a key risk factor. (3) However, as pediatric populations were excluded from this study, these findings cannot be directly extrapolated to children. There is a clear need for prospective, pediatric-specific global databases to better define prognostic markers and optimize long-term management strategies.

<u>Conclusion:</u> This case highlights that ocular myasthenia gravis should be considered even in the absence of positive immunological findings and negative genetics, as early initiation of appropriate therapy may lead to clinical improvement. Furthermore, it underlines the need to collect more pediatric cases to better understand ocular myasthenia in young children and to improve diagnostic strategies and prognostic assessment.

References:

1. Finnis MF, Jayawant S. Juvenile myasthenia gravis: a paediatric perspective. Autoimmune Dis 2011;2011:404101.

2. Case Reports: Juvenile ocular myasthenia gravis: a report of two cases; Ryan Gabbard et al. ;University of South Carolina / Prisma Health, Columbia, South Carolina. Digital Journal of Ophthalmology, Vol. 30, 2023

3. Clinical Characteristics Associated With Secondary Generalization in Patients With Ocular Myasthenia Gravis. A Systematic Review and Meta-analysis; Clarissa Ern Hui Fang et al. ; Neurology® 2023;101:e1594-e1605. doi:10.1212/WNL.0000000000207642

4. Perplexity, Pubmed., aha.scientificposters.com

5. Intern Diagnostics and KISIM

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An uncommon association of anti-NXP2 positive autoimmune polymyositis in a child with localised scleroderma

Jeanclaude Sophie [1] *(sophie.jeanclaude@chuv.ch)*, Cahani Edona [1], Favez Alice [1], Natoli Valentina [1], Arlabosse Tiphaine [1], Armengaud Jean-Baptiste [1]
[1] Division of Pediatrics, Department Women-Mother-Child, Lausanne University Hospital and University of Lausanne, Lausanne, Switzerland.

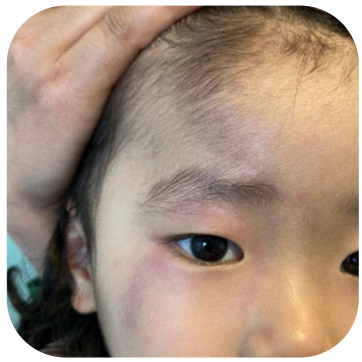
Background
Juvenile localised scleroderma (morphea) :
- Seldom but predominant form of scleroderma in children
- Rarely associated with systemic disease

Autoimmune juvenile polymyositis :
- Rare inflammatory myopathy
- Clinical presentation : proximal muscle weakness and fatigue without skin disease ; may lead to pulmonary or gastrointestinal complications
- Diagnosis : clinical scores, MRI, muscle biopsy and auto-antibody testing
- Treatment : guided by severity ; high-dose glucocorticoids combined with glucocorticoid-sparing immunosuppressive therapy

Case report :
- 5 years-old girl with a rare combination of localized scleroderma and severe auto-immune polymyositis

06.2023
3 y.o.

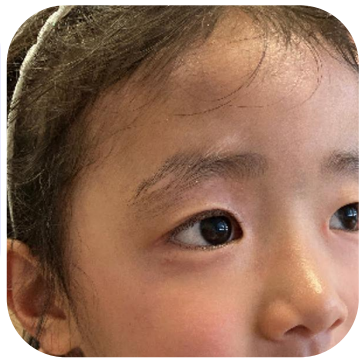
Localised scleroderma «en coup de sabre»
- Oral Methotrexate

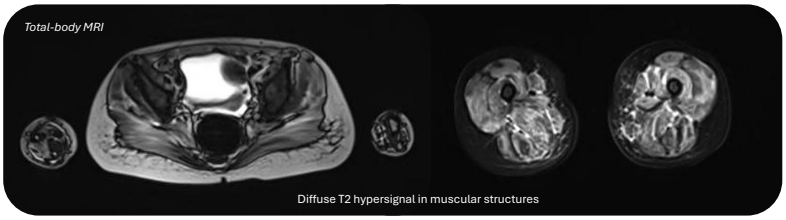


10.2025
5 y.o.

Localised scleroderma «en coup de sabre» : no more cutaneous stigma under Methotrexate treatment

Apparition of progressive lower limb pain and severe muscle weakness affecting daily activities
- Marked proximal weakness (C-MAS 30/52, MMT8 50/80) ; positive Gower's sign ; diffuse muscle tenderness
- No skin or nailfold changes

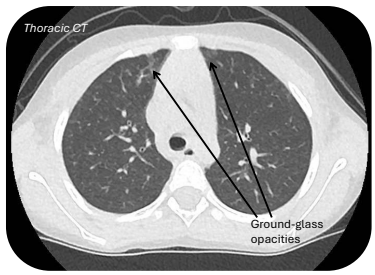




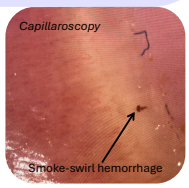
Diagnostic work-up
- Blood work : elevated muscle enzymes (CK 2300 U/L), slightly elevated liver function, mild elevation of inflammatory markers
- Auto-antibody testing : positive anti-NXP2 (associated with severe muscle disease) and anti-PM/Scl100 antibodies (associated with overlap syndrome involving inflammatory myopathies and systemic sclerosis)
- Total-body MRI : severe, diffuse and symmetrical polymyositis
- Muscle biopsy : minimal inflammatory changes
- Capillaroscopy : pathological modifications
- Exclusion of differential diagnosis & Genetic testing

Extension evaluation : pulmonary assessment (normal pulmonary functions, ground-glass opacities on CT) ; cardiac assessment (normal) ; GI assessment (oro-pharyngeal dysphagia, no gastro-intestinal involvement) ; diminished vaccinal response

12.2025
1st hospitalisation (2 weeks long)



Treatment
- Glucocorticoids : multiple doses of high-dose intravenous Methylprednisone followed by oral Prednisone
- Intravenous immunoglobulins
- Rituximab : 1x/week, intravenous
- Replacement of Methotrexate by Mycophenolate Mofetil



Initial outcome
- Treatment is well tolerated
- Good clinical and biological response

01.2026
2nd hospitalisation (1 month long)

Clinical and biological relapse
Treatment changes
- Glucocorticoids : multiple doses of high-dose then normal-dose intravenous Methylprednisone followed by oral Prednisone
- Intravenous Immunoglobulins : 2 doses then once every 2 weeks
- Continuation of Mycophenolate Mofetil and Rituximab
- Addition of oral Ruxolitinib (anti-JAK)
- Infectious prophylaxis : oral Sulfamethoxazole-Trimethoprim, oral Valaciclovir, CMV PCR 1x every 2 weeks

Clinical and biological improvement : augmentation of strength, diminution of dysphagia, stable low CK
- Intravenous Immunoglobulins spaced out to 1x/month and Rituximab to every 6 months ; projected oral Prednisone tapering
- Back to school

But subsequent
- CMV reactivation : introduction of therapeutic Valaciclovir (digestive side effects)
- Corticosteroids side effects : arterial hypertension (oral Amlodipine), oral candidiasis (oral Mycostatin), irritability, cushingoid facies, hairiness

04.2026
6 y.o. – latest out-patient check-up

To be continued

Conclusion
- Extremely uncommon association between localised scleroderma "en coup de sabre" and polymyositis
- Severe muscle involvement without cutaneous changes

→ Extensive evaluation

- Early aggressive high-dose glucocorticoids combined with multiple non-glucocorticoid immunosuppressive treatment

→ Favourable evolution so far (since introduction of anti-JAK!) but subsequent treatment side effects

Kawasaki Disease Shock Syndrome

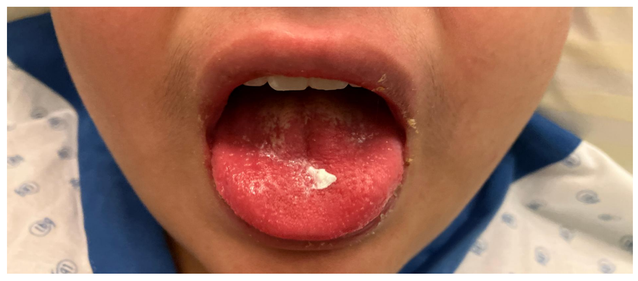
Ravidà H[1], Volery M[1], Lecarpentier J[2], Cahani E[3], Natoli V[3], Armengaud JB[1]

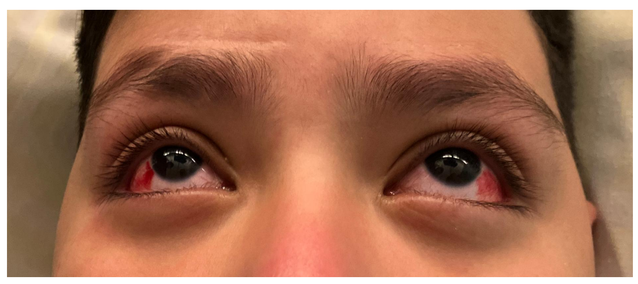
[1]Division of Pediatrics, Department of Women-Mother-Child, Lausanne University Hospital, Switzerland; [2]Pediatric Intensive Care Unit, Department of Women-Mother-Child, Lausanne University Hospital, Switzerland; [3]Pediatric Immunology, Allergology and Rheumatology Unit, Department of Women-Mother-Child, Lausanne University Hospital, Switzerland

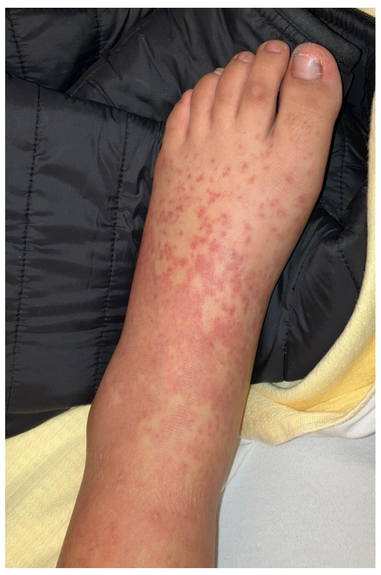
hugo.ravida@chuv.ch

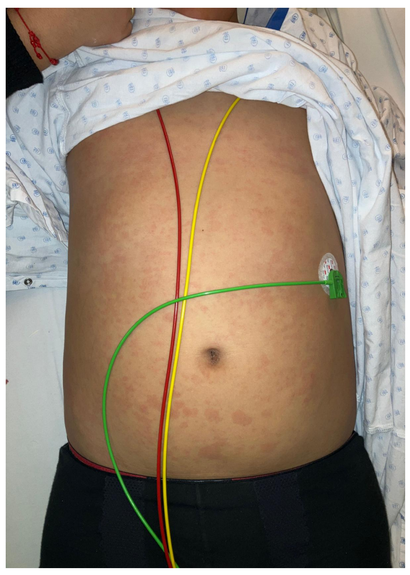
LEARNING OBJECTIVES

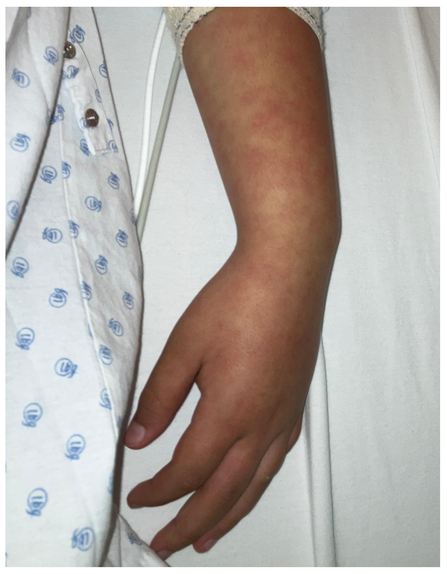
- **Identify** the clinical features of Kawasaki disease shock syndrome
- **Differentiate** Kawasaki disease shock syndrome from other causes of distributive shock and multi-systemic failure
- **Understand** the importance of prompt treatment initiation in order to prevent complications and optimize clinical outcomes

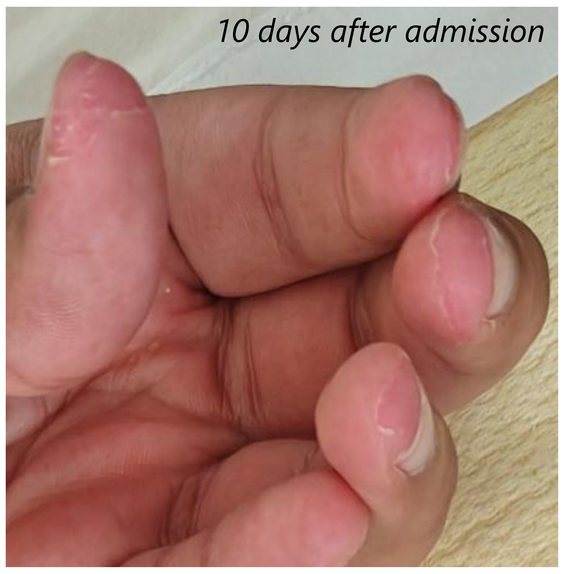












CASE REPORT

- **11 years old boy** from Ecuador, with history of exogenous obesity with hepatic steatosis and asthma
- 4-day fever, with complete **Kawasaki** disease features, signs of **shock** with hemodynamic instability with hypotension and poor peripheric perfusion
- Elevated inflammatory markers (CRP, ESR), hyperferritinemia, normal hemoglobin, normal leukocyte count with high % of neutrophils, thrombocytopenia, hyponatremia, hypoalbuminemia, ASAT ↑, ALAT ↑, creatinine ↑, NT-ProBNP ↑, troponin ↑, normal triglycerides, normal fibrinogen, normal cerebro-thoraco-abdominal CT
- Hemodynamic stabilization obtained with fluid resuscitation alone ; **intravenous immunoglobulin**, **corticosteroids**, anti-inflammatory dose **acetylsalicylic acid** and antibiotic treatment with Ceftriaxone were immediately started ; a single dose of IV albumin was administered
- Clinical and biological response was excellent within the first 48 hours ; antibiotic treatment was stopped after 48 hours and negative blood and urine cultures
- Immediate and long-term cardiac ultrasound showed **no coronary involvement** and a normal myocardial function ; Sars-CoV-2 serologies were negative

ABOUT KAWASAKI DISEASE…

- **Kawasaki disease** is an idiopathic vasculitis of early childhood (**<5 years old**), involving small and medium-sized arteries with tropism for coronary arteries
- Symptoms of fever, polymorphous skin rash, conjunctival hyperemia, erythema of the lips and oral cavity, changes in the extremities, and lymphadenopathy
- If untreated, **15-25%** of children develop coronary artery aneurysm or dilation
- Prompt initiation of therapy with **intravenous immunoglobulin and acetylsalicylic acid** reduces the incidence of coronary artery complications

…AND THE KAWASAKI SHOCK SYNDROME

- **1-5%** of patients with Kawasaki disease present with **hemodynamic instability**
- Affects predominantly **older children** (median ~5 years old)
- Associated with higher risk of **IVIG resistance** and **cardiac involvement**
- Suspected etiology : more intense vasculitis with capillary leak and increased release of cytokines with myocardial dysfunction
- **Early recognition** and distinction from differential diagnosis is key to treat and prevent complications

DIFFERENTIAL DIAGNOSIS

KDSS
Kawasaki disease shock syndrome

- Age : median **5** years old
- **~1-5%** of KD, **incomplete** > complete, digestive symptoms ↑
- Treatment : corticosteroids, IVIG, acetylsalicylic acid
- **Coronary** complications

MAS
Macrophage activation syndrome

- Age : median **5** years old
- **~1%** of KD, **~10%** of sJIA
- Ferritin ↑, platelet ↓, ASAT ↑, triglycerides ↑, fibrinogen ↓
- Treatment : corticosteroids **IVIG resistance ++**
- 25% **mortality** if untreated

TSS
Toxic shock syndrome

- **All ages**
- Diffuse rash, palmoplantar desquamation
- Mainly **toxin**-producing *Streptococcus pyogenes* and *Staphylococcus aureus*
- Treatment of shock and **antimicrobial**

SS
Septic shock

- **All ages**
- Few or absent clinical signs of KD, **infection source signs**
- Shorter duration of fever before admission
- Treatment of shock and **antimicrobial**

MIS-C
Multisystem inflammatory syndrome

- Age : median **6-11** years old
- **KD-like** signs and symptoms
- 0.4–5.5/100'000 children **after COVID-19** infection
- Treatment : corticosteroids, IVIG, acetylsalicylic acid
- **Myocardial** complications

CONCLUSIONS

- **Kawasaki disease shock syndrome** is an entity characterized by hemodynamic instability in the setting of Kawasaki disease
- **Clinical overlap** with macrophage activation syndrome, toxic shock, septic shock or MIS-C makes diagnosis challenging
- **Association with coronary artery complications** requires a prompt treatment (IVIG, corticosteroids, acetylsalicylic acid)

BIBLIOGRAPHY

Gitelman syndrome: a case report

Dr. Med. C. Deli [1] , Dr. Med. S. Franchi [1], Prof. Dr. Med. G. Simonetti [1]
[1]Istituto Pediatrico della Svizzera Italiana, EOC, Bellinzona, Switzerland

Background

Gitelman syndrome is an **autosomal recessive** renal tubulopathy causing **impaired NaCl reabsorption** in the distal convoluted tubule, characterized by hypokalemia, hypomagnesemia, metabolic alkalosis and hypocalciuria. Symptoms can appear during adolescence or adulthood and are often mild, including **muscle weakness, cramps, fatigue, and salt craving,** but they can affect **growth** in children. Diagnosis is based on electrolyte imbalances and **genetic testing**, and treatment with supplements and diet adjustments usually ensures a good prognosis.

Key features:

- Hypokalemia
- Hypomagnesemia
- Metabolic alkalosis
- Hypocalciuria

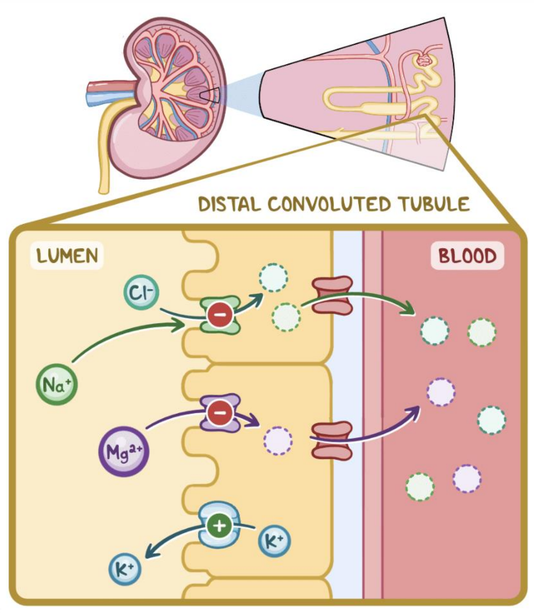


Fig.1: impaired NaCl reabsorption in the distal convoluted tubule [1]

Case

7-year-old healthy girl with 2-day fever and vomiting, history of salty food preference and calf cramps. No family history of renal disease. Physical examination and vitals were normal. Labs showed mild **metabolic alkalosis** (pH 7.47), **moderate hypokalemia** (K^+ 2.8 mmol/L), **hypomagnesemia** (0.65 mmol/L), **hypophosphatemia** (0.92 mmol/L) **and hypocalciuria**. Transtubular potassium gradient (TTKG) 13 indicated renal K^+ wasting. ECG was normal. Renal ultrasound unremarkable, **negative for nephrocalcinosis.** Oral supplementation with K^+ and Mg^{2+} was started. Findings suggested congenital renal tubulopathy, likely Gitelman syndrome.
Genetic testing revealed a mutation in the SLC12A3 gene, confirming the suspicion of Gitelman syndrome.

Labs:

- Metabolic alkalosis pH 7.47
- K^+ 2.8 mmol/L (moderate)
- Mg^{2+} ↓
- Phosphate ↓
- Hypocalciuria

SLC12A3 mutation → Gitelman confirmed

Discussion

Gitelman syndrome can be mild or even undiagnosed for many years. Thorough clinical history and assessment are crucial for raising suspicion. Proper management is key for optimal growth. At the first visit, the patient's weight was in the 4th percentile. Within 25 days of oral supplementation, her weight increased by 900g, reaching the 8th percentile. Muscle cramps disappeared.

Oral K^+ + Mg^{2+} supplementation → **After 25 days:**

- +900 g weight
- 4th → 8th percentile
- Cramps resolved

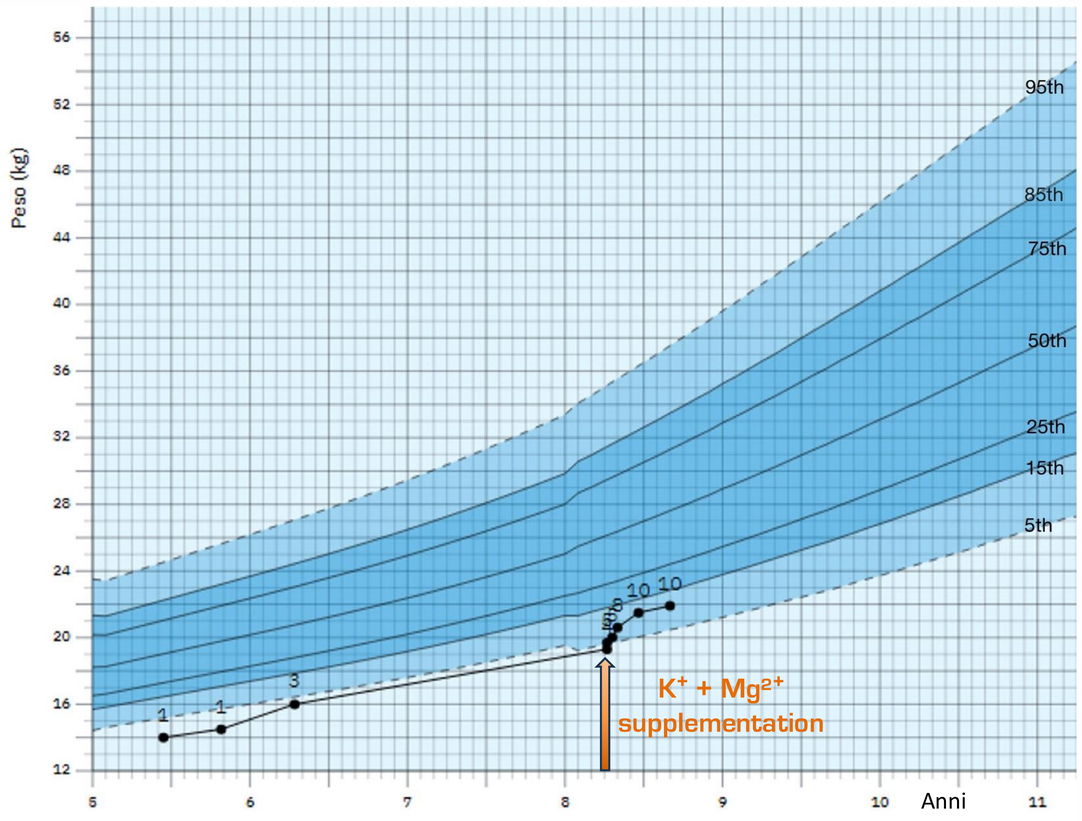


Fig.2: Weight growth curve: from 4° to 8° percentile [2]

Conclusion

Renal tubulopathy should be considered in children presenting with:

- **hypokalemia**
- **metabolic alkalosis**
- **K^+ and Cl^- wasting in urine**

Early recognition and oral supplementation can enhance growth.

References:
1. https://www.osmosis.org/answers/Gitelman-syndrome: Gitelman syndrome: what it is, cause, treatment and more. Author: G. Tiarks,
2. Weight growth curve of patient , 7years old, from August 2025. GECO, EOC.

Not just a hematoma: persistent post-traumatic cranial swelling revealing pediatric Langerhans cell histiocytosis

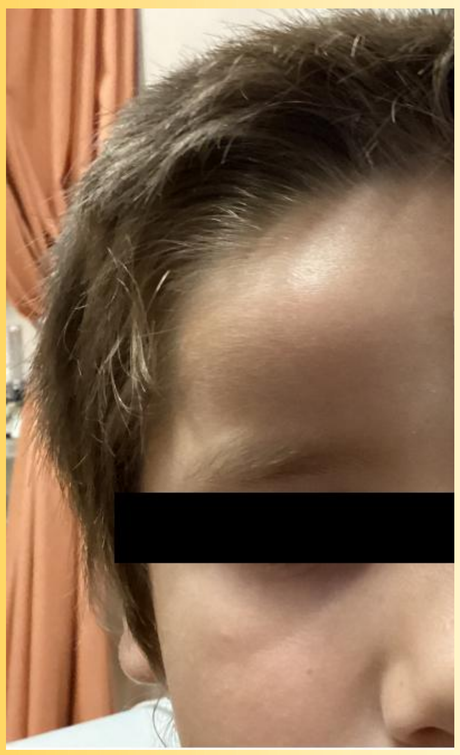
Prochilo A.[1], Proserpio M.[1], Rizzi M.[2], Brazzola P.[2];
[1]Istituto Pediatrico della Svizzera Italiana, OCL, Lugano, Switzerland;
[2]Istituto Pediatrico della Svizzera Italiana, ORBV, Bellinzona, Switzerland.

Background

- Rare clonal disorder of myeloid dendritic cells
- Incidence: 4.6–9 / 1,000,000 children <15 yrs/year
- May mimic benign solitary bone lesions
- Delayed diagnosis: risk of destructive/multisystem disease

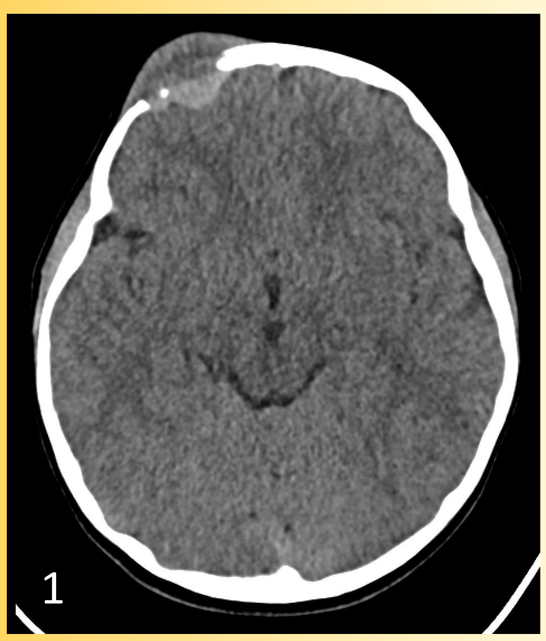
Case

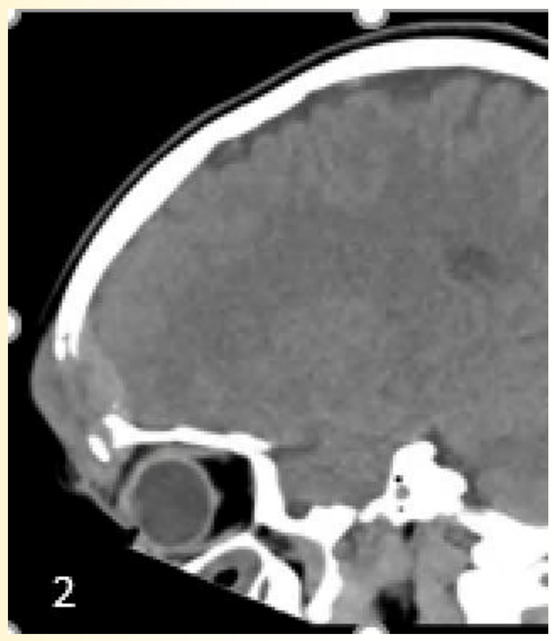
- 5-year-old boy
- Frontal swelling immediate after minor head trauma; unchanged over 2 weeks
- Exam: firm, tender, non-reducible swelling fixed to bone; mild ipsilateral periocular edema; skin erythema
- No fever, vomiting, visual/neurological symptoms; normal neuro & abdominal exam

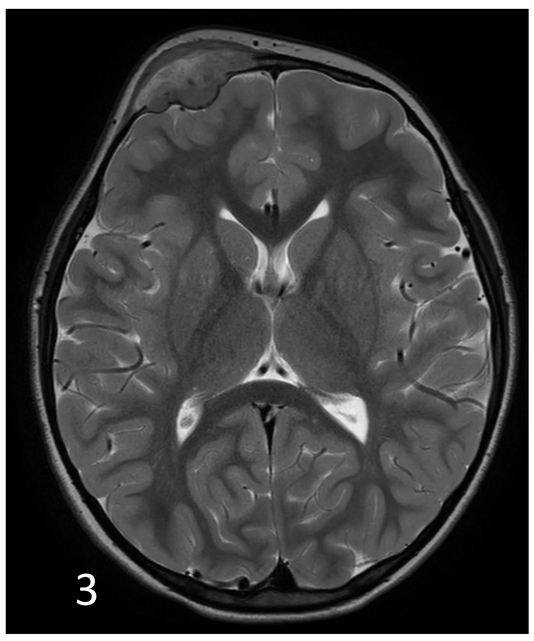


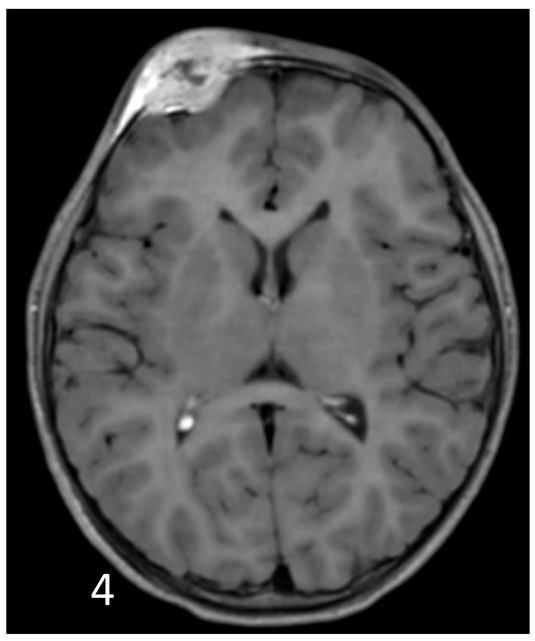
Diagnosis, management and outcome

- **Ultrasound:** Hyperechoic lesion 40×20 mm; outer skull disruption; intracranial extension with focal brain compression
- **CT / MRI:** Osteolytic lesion with soft-tissue extension
- **Histopathology:** LCH confirmed (CD1a+, Langerin+)
- **Treatment:** LCH-IV protocol (stratum I, group 2): IV vinblastine + oral prednisone
- **Evolution**: complete regression without subsequent relapses





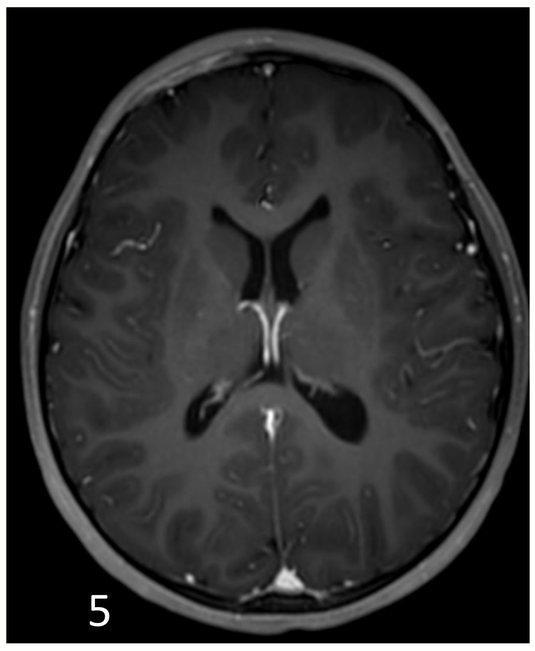




At diagnosis: CT axial and sagittal (1, 2); MRI T2 and T1 with contrast (3, 4)
At 6-month treatment follow-up: MRI T1 (5)

Discussion

- Persistent, firm, unchanged post-traumatic swelling warrants workup for rare pathologies (e.g. LCH)
- LCH skull lesion: consider in differential of cranial swelling after minor trauma, even without systemic symptoms
- Early imaging + histological confirmation ⟶ timely multidisciplinary management



IMMUNE THROMBOCYTOPENIA FOLLOWING MMR VACCINATION IN AN ADOLESCENT GIRL: A Case Report.

Sara E[1], Zwingli G[1], Laoudi Y[1], Schifferli A[2]

[1]Department of Pediatrics, Jura Hospital, Delémont, Switzerland

[2]Department of Hematology and Oncology, University Children's Hospital Basel, Switzerland

1 BACKGROUND

Immune thrombocytopenic purpura (ITP) is an autoimmune disorder characterized by isolated thrombocytopenia due to immune-mediated platelet destruction. In children, ITP commonly follows viral infections. A temporal association between measles–mumps–rubella (MMR) vaccination and ITP has been well documented in the literature [1].

2 CASE REPORT

We report a **12-year-old girl** with a history of recurrent epistaxis and menorrhagia since menarche. One week after receiving the MMR vaccine, she developed daily epistaxis with significant blood loss and gingival bleeding during toothbrushing.

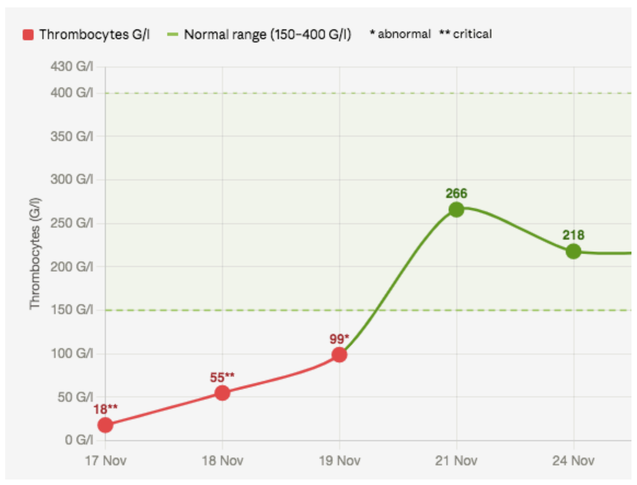


Figure 1. Platelet count evolution during hospitalization.

Laboratory tests showed **severe thrombocytopenia (platelets 18×10⁹/L)**, transient leukopenia, with normal haemoglobin. Inflammatory and haemostasis parameters were within normal limits.

Serologies for CMV, parvovirus B19, EBV, and hepatitis viruses were all negative. Peripheral blood smear and chest radiography excluded haematologic malignancy. Given a maternal history of systemic lupus erythematosus, a complete autoimmune screening was performed and returned negative. Urinalysis was normal.

All infectious, malignant, and systemic autoimmune causes were systematically excluded, and a **diagnosis of post-vaccination ITP associated with MMR immunization** was established.

3 DISCUSSION

Day 0	MMR vaccine administered
Day +7	Daily epistaxis and gingival bleeding onset
Day +10	Severe thrombocytopenia (30×10⁹/L) confirmed
Day +13	Secondary causes excluded — ITP diagnosed
Follow-up	Clinical improvement under treatment

ITP is the most common acquired bleeding disorder in children and is triggered by immune stimulation. Post-vaccination ITP is rare, but a significant risk increase has been documented within 6 weeks of MMR vaccination [1]. Molecular mimicry and immune dysregulation are thought to lead to the production of antiplatelet autoantibodies.

Clinically, patients present with mucocutaneous bleeding of variable severity. Diagnosis is clinical, based on isolated thrombocytopenia after careful exclusion of secondary causes. In this case, the normal inflammatory and haemostasis markers, negative infectious serologies, and negative autoimmune panel all supported a primary ITP diagnosis, with the recent MMR vaccination as the most probable triggering event.

Most paediatric cases of post-MMR vaccination ITP are benign and self-limited, and respond well to standard therapy when clinically indicated. Early recognition and appropriate management are essential to avoid unnecessary investigations and to ensure the continued safety of vaccination programmes [2].

4 CONCLUSION

Post-MMR vaccination ITP is a rare but well-recognized condition that should be considered in any child presenting with acute thrombocytopenia and mucosal bleeding following recent immunization. Heightened awareness of this association supports prompt and accurate diagnosis, guides appropriate clinical management, and should not deter clinicians from continuing to recommend vaccination according to established schedules [2].

Post-MMR ITP: rare but important — always consider in acute thrombocytopenia post-vaccination.

5 REFERENCES

[1] Black C et al. MMR vaccine and idiopathic thrombocytopaenic purpura. Br J Clin Pharmacol. 2003;55(1):107–111.

[2] Abik M, et al. Severe immune thrombocytopenia following MMR vaccination with rapid recovery: A case report and review of literature. Int Med Case Rep J. 2020;13:697–699.

Pädiatrie Schweiz · 11–12 June 2026

Département femme-mère-enfant
Pneumologie et mucoviscidose

The Mystery of the Vanishing Lung:

Swyer-James Syndrome Following Suspected COVID-19 in a Child

T. Bernasconi[1] M. Franchina[1] C. Fernandez Elviro[1] S. Blanchon[1] E. Tenisch[2] I. Rochat[1]

[1] Paediatric Pulmonology & Cystic Fibrosis Unit, Dept. Femme Mère Enfant – CHUV, Lausanne | [2] Radiodiagnostic & Interventional Radiology – CHUV, Lausanne

CLINICAL TIMELINE

Nov 2020 · Age 3y — COVID-19 pneumonia
1 month hospitalisation

2021–2023 · Age 4–6y — ~1 pneumonia/year
Wheezing · exertional dyspnoea

2024 · Age 7y — Dyspnoea on exertion
Oral corticosteroids trial
→ no improvement

Jun 2024 — Chest X-Ray
Left hyperlucency

Aug 2024 — Chest CT
Diagnosis of SJS

2024–2025 · Now — Infection-free
improved Spirometry

BACKGROUND & CASE

Patient — 7-year-old male

Nov 2020 (age 3y): severe COVID-19 pneumonia, 1 month hospital stay

2021–2024 : Recurrent infections, intermittent wheezing, progressive exertional dyspnea

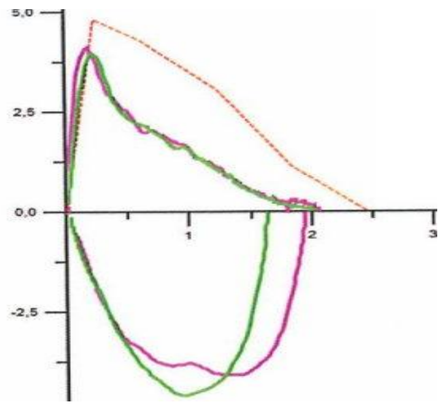
Clinical exam: asymmetric auscultation with **reduced breath sounds left**.

Spirometry : **Obstructive pattern, no bronchodilator response (FEV1 +1.9%) - *fixed obstruction***

Treatment trials :

- Inhaled corticosteroids → partial response only, no resolution of fixed obstruction
- Oral corticosteroids → no improvement of fixed obstruction

SPIROMETRY



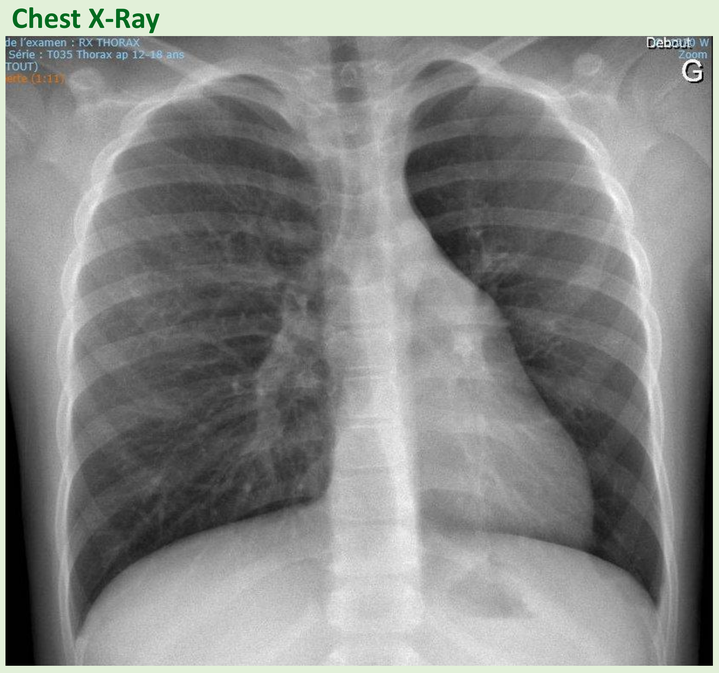
74%
FEV1/FVC
z-score −1.82

73%
FEV1
z-score −2.32

49%
DEM 25-75
z-score −2.55

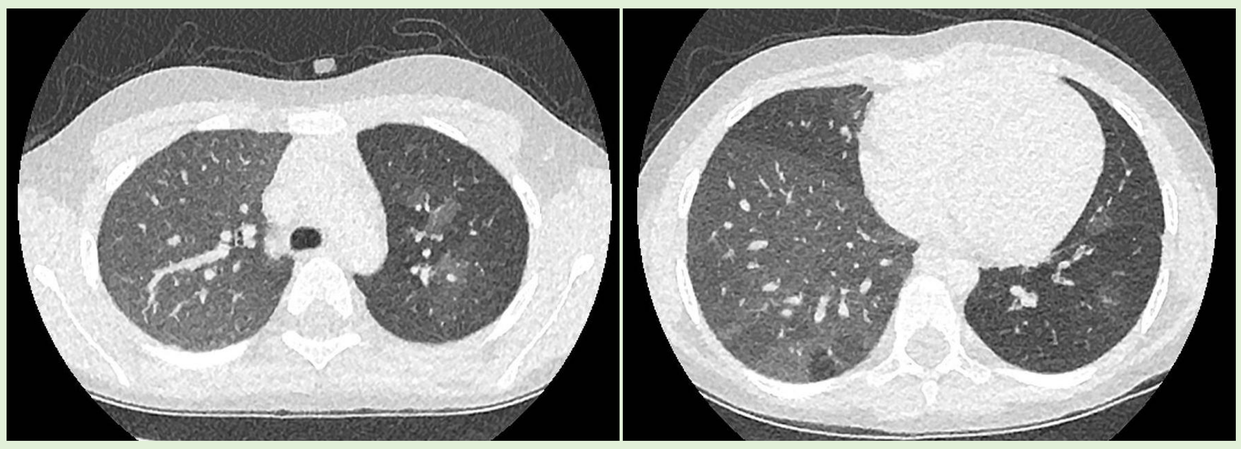
RADIOLOGICAL FINDINGS — Key Diagnostic Evidence

Chest X-Ray

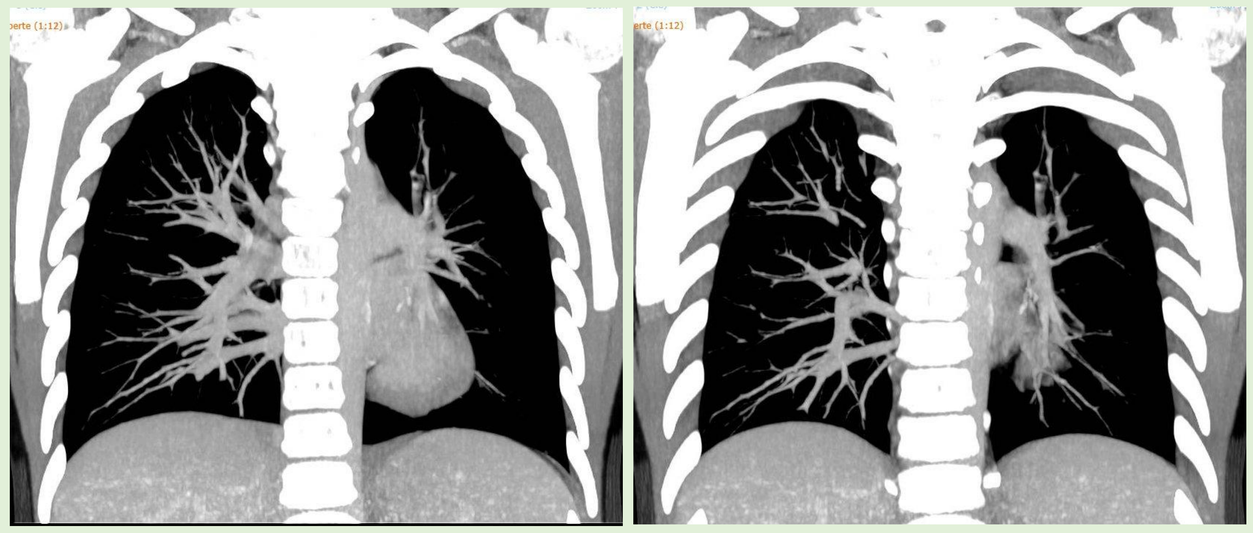


Right-to-left asymmetry :
increased left lung transparency

Chest CT



Mosaic attenuation pattern :
diffuse hypodensity of the left parenchyma, air trapping



Vascular rarefaction :
Decreased vascularity in the left lobe

DISCUSSION

Swyer-James Syndrome (SJS)

Rare post-infectious obliterative bronchiolitis (= inflammatory injury to the small airways, leading to submucosal fibrosis and partial or complete luminal obliteration) localized in one lobe → unilateral hyperlucent lung.
Most common causative agent: **Adenovirus**.

Diagnostic SJS

- Imaging: unilateral hyperlucency, vascular rarefaction + mosaic CT pattern
- Spirometry: fixed obstruction
- **History:** severe respiratory infection in early childhood
- **Exclusion** of alternative diagnoses (pneumothorax, foreign body, congenital malformation, immunodeficiency
- **Why SJS and not asthma?** Fixed obstruction — no bronchodilator response in spirometry and no benefit from oral corticosteroid trial

Why COVID-19 as causative agent?

SARS-CoV-2 causes diffuse alveolar damage, bronchiolar inflammation and organising pneumonia - the same end pathway as obliterative bronchiolitis. Severe COVID-19 at age 3, during a critical lung development window, likely amplified irreversible airway damage.
To our knowledge, **no prior case identifies COVID-19 as causative factor of SJS in a child.** Post-pandemic surveillance for long-term pulmonary sequelae in children is essential.

PATHOPHYSIOLOGY OF SWYER-JAMES SYNDROME

Severe respiratory infection
Distal obstruction → hyperinflation

Granulation tissue proliferation
Bronchiolar inflammation

Small airway obliteration
→ Fibrosis + alveolar damage

Air trapping
Distal to obstruction -> hyperinflation

Fibrotic septa
compress capillaries

↑ Capillary resistance
vascular remodelling

Compensatory vasoconstriction
due to hypoventilation

Unilateral lung hyperinflation
Air trapping → hyperlucency on CXR

Unilateral hypoperfusion
Mosaic pattern + ↓ vessel calibre on CT

Swyer-James Syndrome — Unilateral hyperlucent lung

CXR: left hyperlucency + mediastinal shift · CT: mosaic perfusion + air trapping
Spirometry: fixed obstruction · no BD response · LCI elevated

CONCLUSION

Diagnosis

Unilateral hyperlucency on CXR + mosaic perfusion + expiratory air trapping on CT are key imaging findings. Fixed obstruction with no BD response on spirometry + elevated LCI confirm diagnosis of SJS.

COVID-19 as cause

First reported paediatric case of SJS following COVID-19.
Post-pandemic surveillance for long-term pulmonary sequelae in children is essential.

Targeted therapy

High-dose systemic CS do not benefit fixed obstruction.
Targeted approach: ICS + seasonal azithromycin + mucolytics + early antibiotic therapy at infection + pneumococcal and influenza vaccinations

REFERENCES

1. Behrendt A, Lee Y. Swyer-James-MacLeod Syndrome. StatPearls. StatPearls Publishing; 2025.
2. Chang AB et al. ERS guidelines for management of children with bronchiectasis. Eur Respir J. 2021;58(2):2002990.
3. Li AM et al. Non-CF bronchiectasis: does knowing the aetiology change management? Eur Respir J. 2005;26(1):8-14.
4. Sen HS et al. Adult diagnosis of Swyer-James-Macleod syndrome. Respir Care. 2014;59(4):e51-4.
5. Sulaiman A et al. Swyer-James-MacLeod syndrome misdiagnosed as pneumothorax. Interact Cardiovasc Thorac Surg. 2009;8(4):482-4.
6. Swyer PR, James GC. A case of unilateral pulmonary emphysema. Thorax. 1953;8(2):133-6.
7. Chaucer B et al. Swyer-James-Macleod syndrome in the ED. Am J Emerg Med. 2016;34(7):1329.e3-4.

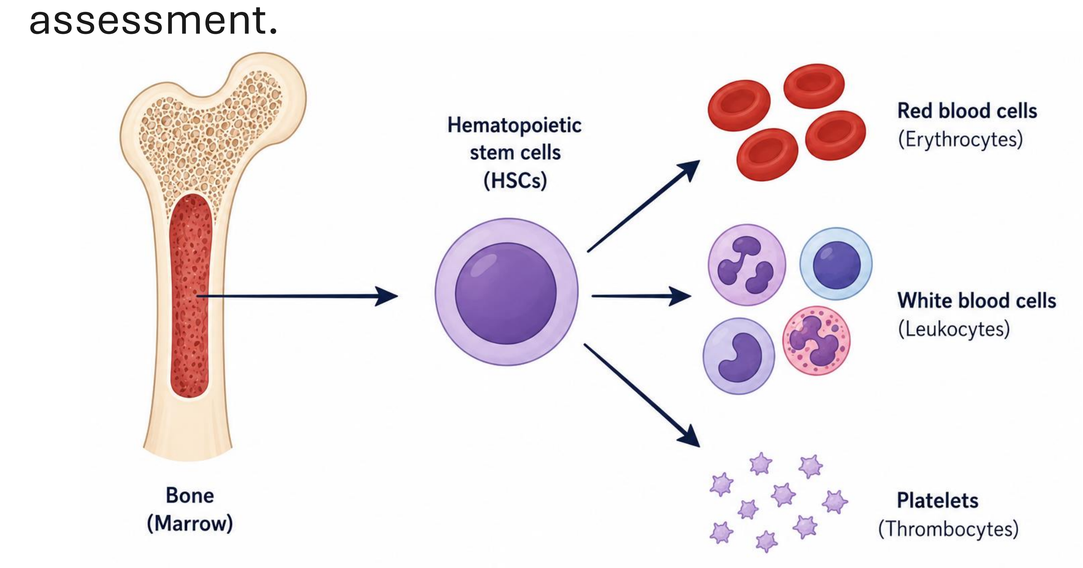
Severe pancytopenia in a child with trisomy 21: a challenging differential diagnosis

Roxane Zweifel[1], Valentin Tammaro[1], Giovanna Alfano[2], Manuel Diezi[3], Bastien Guggisberg[1], Renato Gualtieri[1]

1. Division of Pediatrics, Department of Women-Mother-Child, Lausanne University Hospital and University of Lausanne, Lausanne, Switzerland. 2. Gastro-enterology Unit, Division of Pediatrics, Department of Women-Mother-Child, Lausanne University Hospital and University of Lausanne, Lausanne, Switzerland. 3. Hematology and oncology Unit, Division of Pediatrics, Department of Women-Mother-Child, Lausanne University Hospital and University of Lausanne, Lausanne, Switzerland

Introduction

- Pancytopenia in children covers a broad differential, from transient and reversible causes to bone marrow failure and hematological malignancies.
- Clinical presentation is often non-specific; comorbidities and communication difficulties further complicate assessment.



Case presentation

Patient: 15-year-old girl with trisomy 21 and autism spectrum disorder.

History: 3 months of progressive asthenia and anorexia, highly selective eating, 2-kg weight loss over one year. No diarrhea or other digestive symptoms.

Examination: pallor, tachycardia, new 2/6 systolic murmur.

Laboratory:

- Severe macrocytic anemia — Hb 61 g/L, MCV 126 fL, low reticulocytes
- Neutropenia 0.89 G/L; thrombocytopenia 33 G/L
- Elevated LDH; subclinical hypothyroidism

Differential diagnosis & workup

Hypothesis	Findings
Malignancy (leukemia)	Smear: no blasts. Bone marrow & flow cytometry: no blasts; megaloblastic pattern compatible with B12 deficiency.
Nutritional deficiency	Vitamin B12 ↓↓ Folate ↓ Iron normal
Infection	EBV, CMV: immune. Parvovirus B19: negative.
Hemolysis / TLS / Autoimmune	Isolated LDH ↑; direct Coombs negative; no other features.
Bone marrow failure / aplasia	Bone marrow examination: no evidence.

Workup of B12 deficiency:

- Autoimmune pernicious anemia: negative intrinsic factor and parietal cell antibodies, normal gastroscopy → excluded.
- Nutritional assessment → confirmed a **dietary etiology** with selective eating habits.

Treatment and evolution:

- Red blood cell transfusion + parenteral vitamin B12 supplementation; nutritional follow-up.
- Transient post-supplementation nadir of the other cell lines, followed by rapid and sustained hematological recovery.
- Hb 99 g/L after transfusion → 139 g/L at 2 months, with continued improvement thereafter.

Conclusion:

- Severe vitamin B12 deficiency can present with pancytopenia and bone marrow dysplasia mimicking malignant or inflammatory disease.
- It should be considered in children with neurodevelopmental conditions (e.g. autism spectrum disorder, trisomy 21) and restrictive eating behaviors.
- Beyond hematological changes, B12 deficiency can also cause neurological and developmental complications.
- **Prompt recognition allows simple, effective treatment and rapid recovery.**