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Case Report
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Recurrent Respiratory Tract Infections and Pneumonia in a 4-Year-Old Girl with Unrecognized Hypoganglionosis Successfully Treated with Transanal Endosurgical Modified Anorectal Myomectomy

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Abstract

We report a 4-year-old girl with recurrent respiratory tract infections and multiple episodes of pneumonia. Despite repeated hospital admissions, the underlying cause remained elusive until severe fecal retention was identified as a contributing factor. Further evaluation revealed previously unrecognized hypoganglionosis. Imaging demonstrated massive fecal loading with diaphragmatic elevation. Rectal biopsies confirmed reduced ganglion cell density consistent with hypoganglionosis. The patient underwent transanal endosurgical modified anorectal myomectomy. Postoperatively, she experienced complete resolution of constipation, improved respiratory mechanics, and cessation of recurrent infections on long term follow up.

Keywords: Recurrent Respiratory Tract Infections; Pneumonia; Pediatric Hypoganglionosis; Chronic Constipation; Malnutrition; Aspiration Pneumonia; Gastrointestinal Motility Disorder; Transanal Endosurgical Approach; Modified Anorectal Myomectomy; Pediatric Surgery

Introduction

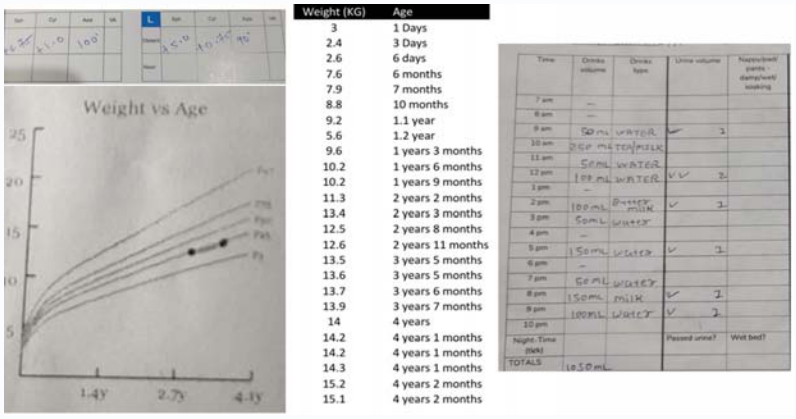
Hypoganglionosis is a rare congenital disorder of the enteric nervous system characterised by reduced density of ganglion cells within the myenteric plexus. It lies within the spectrum of intestinal neuronal dysplasias and may mimic Hirschsprung disease, though its presentation is often more insidious. Children typically present with chronic constipation, abdominal distension, or features of intestinal pseudo-obstruction.

Respiratory complications are not commonly associated with hypoganglionosis. However, severe fecal retention can lead to silent regurgitations during sleep due to reflux following gastric push by splenic flexure, diaphragmatic splinting, impaired lung expansion, basal atelectasis, and increased susceptibility to lower respiratory tract infections. In such cases, the gastrointestinal pathology may remain unrecognised, particularly when respiratory symptoms dominate the clinical picture.

This case underscores the importance of a holistic, cross-system approach to recurrent pediatric infections and demonstrates the effectiveness of transanal endosurgical modified anorectal myomectomy as a minimally invasive, sphincter-preserving treatment option.

Case Presentation

A 4-year-old girl was referred for evaluation of recurrent respiratory tract infections and multiple episodes of pneumonia requiring hospitalisation. Her symptoms began in infancy with bronchiolitis followed by viral induced wheeze, upper respiratory tract infectionsand were characterised by persistent respiratory distress, wheezing, shortness of breath, cough, fever, and reduced exercise tolerance during infectious episodes.



Weight (KG)	Age
3	1 Days
2.4	3 Days
2.6	6 days
7.6	6 months
7.9	7 months
8.8	10 months
9.2	1.1 year
5.6	1.2 year
9.6	1 years 3 months
10.2	1 years 6 months
10.2	1 years 9 months
11.3	2 years 2 months
13.4	2 years 3 months
12.5	2 years 8 months
12.6	2 years 11 months
13.5	3 years 5 months
13.6	3 years 5 months
13.7	3 years 6 months
13.9	3 years 7 months
14	4 years
14.2	4 years 1 months
14.2	4 years 1 months
14.3	4 years 1 months
15.2	4 years 2 months
15.1	4 years 2 months

Figure 1: Clinical Charts.
A. Weight Vs Age growth chart showing twenty-five percentile B. Eyesight tests results, C full record of the weight at various ages with failure to thrive and significant weight loss at 1.2 years due to severe attack of an enterocolitis which required admission and iv antibiotics with follow up catch up growth on recovery and good recovery of weight with initial conservative management after 4 years of age but then plateaued just before surgery D. Input output chart for monitoringdisease progress and medical therapy.

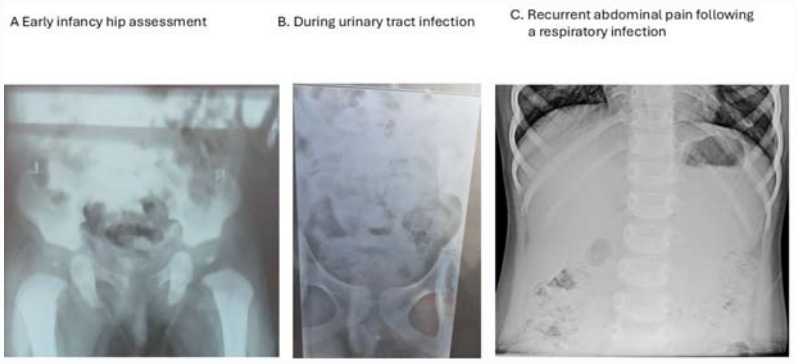


Figure 2: Pelvic and abdominal radiographs from early infancy to 3 years.
A. Bilateral normal hip joints but dilated rectum and sigmoid loaded with fecal matter and gasat 3 months of age B. Post evacuation film of micturating cystourethrogram following an episode of urinary tract infection at the age of 2 years. C. Abdominal radiograph at the follow up of resolved pneumonia at 3 years due to recurrent abdominal pains -note increasing fecal and gas retention in the colon.

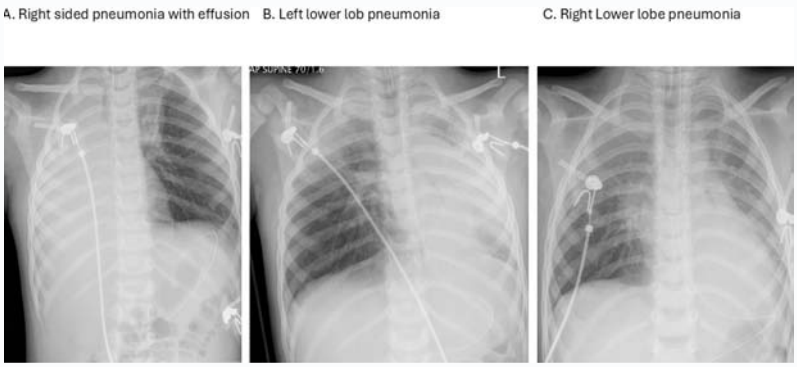


Figure 3: Chest radiographs.
A. first episode with right side pneumonia with collapse consolidation and shift of mediastinum to right and left upper lobar pneumonia with compensatory hyperinflation of the left lung-Note dilated colon in the abdomenB. Second episode with left side pneumonia C. Third episode of right lower lobar apical segmental pneumonia.

Clinical charts showed failure to thrive with growth at 25% centile, high eyesight correction, age-weight chart (note failure to thrive with weight reductions and weight gain after conservative treatment but then lost it due to noncompliance and chose the surgical treatment option) and input output chart (Figure 1).

At the age of 3 months, patient had deep and asymmetric left thigh fold but both hip radiographs and ultrasounds were normal (Figure 2A). She had urinary tract infections for whichpelvic ultrasound and micturating cystourethrogram and postvoid film were all normal (Figure 2B). She then developed recurrent lower respiratory infections

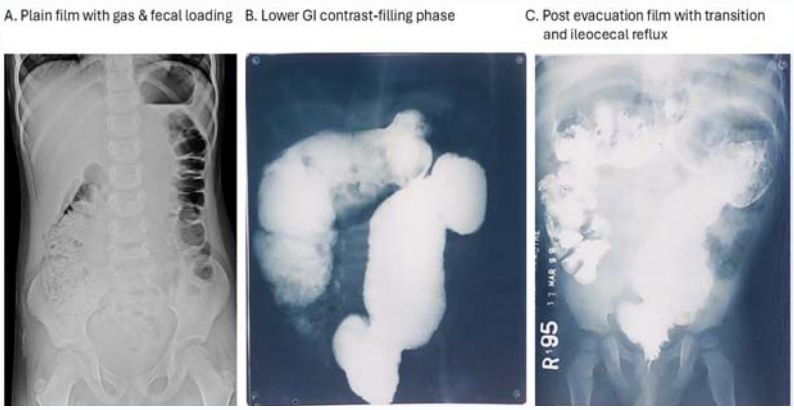


Figure 4: Radiological studies.
A. Plain abdominal film with fecal retained megarectum and rectosigmoid with massive retention of gas in proximal colonB Lower GI Contrast filling phase showing transition at rectosigmoid and proximal hugely dilated colon, appendix and small contrast leaked into terminal ileal loops C Post-evacuation film showing clear transition zone at rectosigmoid, dilated redundant colon loaded with fecal matter mixed with contrast and gas pockets allowing double contrast and free reflux into the terminal ileum.

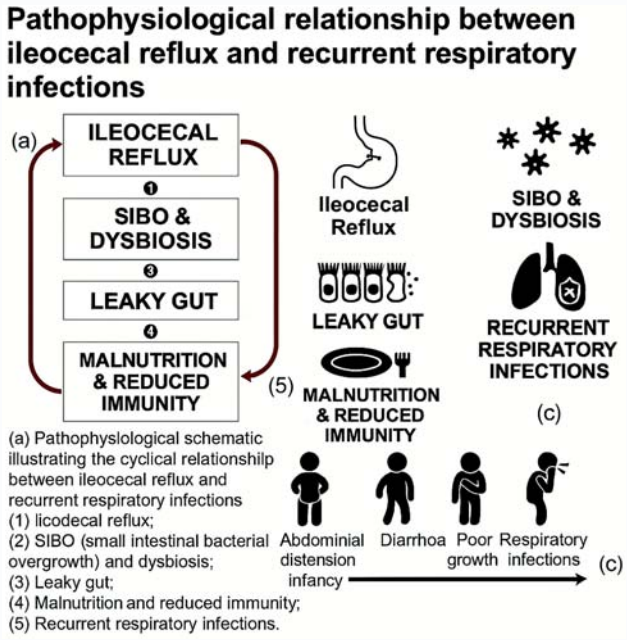


Figure 5: Clinical Pattern: The Gut–Lung–Pneumonia Loop.
1. Colorectal dysmotility + ileocecal reflux → SIBO and dysbiosis
2. Leaky gut + malabsorption → micronutrient deficiency and immune compromise
3. Respiratory vulnerability → recurrent pneumonia
4. Antibiotic use → worsens gut dysbiosis
5. Cycle repeats, deepening systemic dysfunction

requiring three hospital admissions (Figure 2C and Figure 3).

Her parents reported chronic constipation with intermittent self-limiting recurrent enterocolitis in the form of diarrhea and vomiting or overflow diarrheasince early childhood, with infrequent, hard stools and prolonged straining. These symptoms had been attributed to functional constipation and managed intermittently with laxatives for constipation and oral rehydration solutions during diarrhea, with limited benefit. This was considered as functional constipation and was not thought to be causing any apparent symptoms apart from recurrent abdominal pain, worm infestations, vulvovaginitis, labial fusion and urinary tract infections.

On examination, she appeared underweight 25 percentile on growth chart but alert. Her abdomen was markedly distended with palpable fecal masses. Respiratory examination revealed reduced breath sounds at both lung bases. No perianal anomalies or neurological deficits were noted.

Investigations

Laboratory findings showed mild anemia of chronic disease with hemoglobin of 8 gm/dl., normal inflammatory markers between infections and normal immunoglobulin profile. Abdominal radiograph showed extensive rectosigmoid fecal loading with megarectum and plenty of gas in the left, transverse and right colon

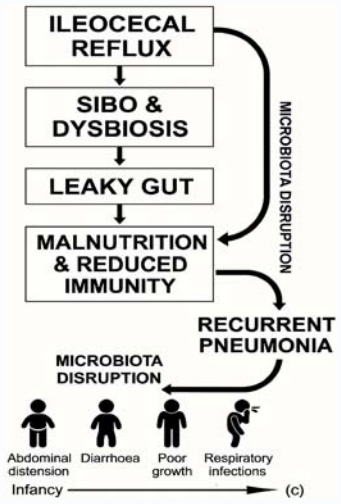


Figure 6: The gut–lung–pneumonia loop.

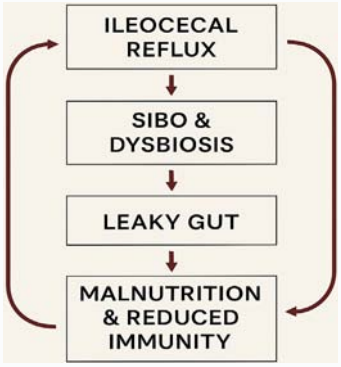


Figure 7: Visual schematic of this gut–lung axis loop

(Figure 3A). Lower gastrointestinal contrast enemaon filling phase showed caliber change at rectosigmoid area suggestive of a distal motility disorder (Figure 3B). The post evacuation film demonstrated plenty of retention of contrast and ileocecal reflux in addition to the transition zone (Figure 3C).

A provisional diagnosis of hypoganglionosis was likely in view of above findings. Differential diagnosis included Hirschsprung disease, internal anal sphincter achalasia, chronic intestinal pseudo-obstruction, functional constipation, cystic fibrosis (due to recurrent pneumonia), primary immunodeficiency.

Initial Clinical and Medical Management

Patient was started on initial conservative management with diet and nutritional optimisation,bowel clean-out with regular laxatives, holobiotics(pre-pro-post-biotics), broad spectrum multivitamin multimineral multi-traceelement supplements with additional iron and calciumsupplements which allowed catch up growth but patient was about to start regular primary school and parents opted for transanal endosurgical correction procedures including full thickness rectal biopsy and modified anorectal internal sphincter and circular smooth muscleextended myomectomy.

Definitive Surgical Intervention

Given the confirmed diagnosis and persistent symptoms, the patient underwent transanal endosurgical modified extended anorectal myomectomy (which is similar to Hellar's cardiomyotomy for achalasia cardia and Ramstedt's pyloromyotomy for infantile hypertrophic pyloric stenosis), a minimally invasive, sphincter-preserving procedure designed to remove the functional

Gut–Lung Axis: The Bridge Between Dysbiosis and Respiratory Vulnerability

Gut Dysfunction	Systemic Consequence	Respiratory Impact
Ileocecal reflux → SIBO → leaky gut	Translocation of microbial products (e.g. LPS, peptidoglycan)	Chronic low-grade inflammation, immune dysregulation
Malabsorption of micronutrients (A, D, E, zinc, selenium, B12)	Impaired mucosal immunity, reduced antibody production	Weak respiratory epithelial defence, poor pathogen clearance
Dysbiosis and reduced SCFA production	Loss of anti-inflammatory tone	Increased susceptibility to viral and bacterial infections
Chronic gut inflammation	Activation of systemic TLRs, cytokine imbalance	Heightened airway reactivity, asthma-like symptoms

Table 1:

Panel (c) Legend: Clinical Timeline of Symptom Progression

This timeline illustrates the typical evolution of symptoms in pediatric patients with colorectal motility disorders and ileocecal reflux:

Age Stage	Symptom	Pathophysiological Link
Infancy	Abdominal distension	Colorectal dysmotility and ileocecal reflux
Early Childhood	Diarrhoea	SIBO and dysbiosis disrupting digestion
Toddler Stage	Poor growth	Malabsorption and micronutrient deficiency
Preschool Age	Fatigue	Immune compromise and systemic inflammation
School Age	Recurrent respiratory infections	Gut–lung axis dysfunction

Table 2:

Mechanistic Bridge: From Gut Dysfunction to Pneumonia

Gut Pathology	Systemic Effect	Pulmonary Consequence
Ileocecal reflux → SIBO	Retrograde bacterial overgrowth	Increased intestinal permeability (leaky gut)
Leaky gut	Translocation of microbial products (e.g. LPS, peptidoglycan)	Systemic inflammation, immune dysregulation
Micronutrient malabsorption (A, D, B12, zinc)	Impaired mucosal immunity, reduced antibody production	Weak respiratory epithelial defence
Altered gut microbiota	Reduced SCFA production, loss of anti-inflammatory tone	Heightened susceptibility to infection
Immune compromise	Blunted innate and adaptive responses	Poor clearance of respiratory pathogens
Antibiotic use for pneumonia	Further gut dysbiosis	Vicious cycle of gut–lung disruption

Table 3:

obstruction by dysfunctional segment and restore normal motility. The operation was completed without complications.

Histopathology

Rectal suction biopsy demonstrated reduced density of ganglion cells, hypoplastic myenteric plexus consistent with features of hypoganglionosis. Supportive immunohistochemistry depicted reduced calretinin staining confirming the final diagnosis.

Outcome and Follow-Up

Postoperatively, the patient demonstrated immediate reduction in abdominal distension and no pain or discomfort anymore, normalisation of stool frequency and consistency, improved respiratory mechanics with full diaphragmatic excursion, no further episodes of pneumonia, improvedappetite, and catch-up growth, improved eye sight with marked reduction in numbers, improved performance in study and concentration. Her quality of life improved significantly, and her parents reported a marked reduction in healthcare utilisationover next3 decades of follow-up.Patient has completed her professional postgraduate education and started her professional career.

Discussion

Hypoganglionosis is a rare congenital disorder of the enteric nervous system characterised by reduced ganglion cell density within the myenteric plexus, leading to impaired colonic motility and chronic constipation. Although gastrointestinal symptoms dominate the clinical picture, extra-intestinal manifestations may occur when severe fecal retention exerts secondary physiological effects. In this case, recurrent respiratory tract infections and pneumonia were the presenting features, overshadowing the underlying motility disorder.

Recurrent respiratory infections are common in early childhood and are often attributed to viral exposures, immature immunity, or benign environmental factors. Most children experience six to ten respiratory infections per year, and the majority do not indicate underlying pathology. However, red-flag features—such as failure to thrive, persistent symptoms, or recurrent pneumonia—warrant broader evaluation for systemic or structural causes. In our patient, repeated lower respiratory tract infections and basal atelectasis prompted further investigation beyond the respiratory system.

The link between severe constipation and respiratory compromise is physiologically plausible. Massive fecal loading can elevate the diaphragm, reduce lung volumes, and predispose to atelectasis and infection. Although this mechanism is well recognised in paediatric practice, it is often under-appreciated in the context of recurrent respiratory disease. The child's chronic constipation had been attributed to functional causes, a common diagnostic pitfall given that most recurrent infections in children are benign and not associated with immune deficiency. This contributed to a delay in recognising the underlying gastrointestinal pathology.

The diagnosis of hypoganglionosis requires a high index of suspicion, particularly when symptoms are atypical. In this case, abdominal distension and megarectum on imaging provided the first clue. Histopathological confirmation remains the gold standard, with reduced ganglion cell density and hypoplastic myenteric plexus supporting the diagnosis. Early recognition is essential, as prolonged fecal retention can lead not only to respiratory complications but also to nutritional compromise, impaired growth, and reduced immunity.

Surgical intervention is the mainstay of treatment for symptomatic hypoganglionosis. The transanal endosurgical modified anorectal myomectomy performed in this case aligns with contemporary minimally invasive approaches used in related motility disorders such as Hirschsprung disease. Transanal techniques have demonstrated favorable functional outcomes, reduced morbidity, and excellent preservation of continent mechanisms in paediatric populations. In our patient, the procedure resulted in rapid decompression, restoration of normal bowel function, and complete resolution of respiratory symptoms—highlighting the interconnectedness of gastrointestinal and respiratory physiology.

This case reinforces several important clinical lessons. First, recurrent respiratory infections, although common—should prompt broader evaluation when accompanied by gastrointestinal symptoms or growth concerns. Second, chronic constipation unresponsive to standard therapy warrants investigation for underlying motility disorders. Finally, minimally invasive transanal surgery offers an effective and durable solution for selected cases of hypoganglionosis, with the potential to resolve both gastrointestinal and secondary systemic manifestations.

Hypoganglionosis is a rare intestinal neuronal disorder typically presenting with chronic constipation, abdominal distension, or pseudo-obstruction. Extra-intestinal manifestations are uncommon and may obscure the underlying diagnosis.

This case illustrates an uncommon but clinically significant interaction between gastrointestinal dysmotility and respiratory pathology. Hypoganglionosis, though rare, should be considered in children with chronic constipation unresponsive to standard therapy. In this patient, massive fecal retention caused diaphragmatic elevation, reducing lung volumes and predisposing her to recurrent infections.

The diagnostic challenge lay in the dominance of respiratory symptoms, which overshadowed the underlying gastrointestinal disorder. This highlights the importance of thorough abdominal assessment in children with recurrent pneumonia, particularly when growth falters or chronic constipation coexists.

Transanal endosurgical modified extended anorectal myomectomy offers a minimally invasive, function-preserving approach with excellent outcomes in selected cases of distal hypoganglionosis. In this patient, the procedure resulted in complete resolution of both gastrointestinal and respiratory symptoms, demonstrating the interconnectedness of organ systems and the value of multidisciplinary evaluation.

This case describes an unusual and clinically significant presentation of pediatric hypoganglionosis manifesting primarily as recurrent respiratory tract infections. The diagnostic pathway highlights how extra-intestinal symptoms may obscure an underlying gastrointestinal motility disorder, leading to repeated hospital admissions and delayed intervention. Our report underscores the importance of maintaining a broad differential diagnosis when evaluating recurrent infections in children, particularly when chronic constipation or abdominal distension coexist.

The case also demonstrates the effectiveness of transanal endosurgical modified anorectal myomectomy as a minimally invasive, sphincter-preserving treatment option. The patient experienced complete resolution of both gastrointestinal and respiratory symptoms following surgery, illustrating the interconnectedness of abdominal and respiratory physiology and the

value of multidisciplinary assessment.

We believe this case will be of interest to clinicians in pediatrics, paediatric surgery, gastroenterology, and general practice, as it provides important insights into atypical presentations of enteric neuromuscular disorders and reinforces the need for early recognition and targeted intervention.

Patient Perspective

The patient's parents described years of distress due to repeated respiratory infections and hospital admissions. They expressed relief at finally receiving a unifying diagnosis and were grateful for the transformative impact of surgery on their daughter's health and wellbeing.

Learning Points

- Hypoganglionosis may present with atypical extra-intestinal manifestations, including recurrent respiratory infections.
- Severe fecal retention can impair diaphragmatic movement and predispose to pneumonia.
- Chronic constipation unresponsive to standard therapy warrants evaluation for underlying motility disorders.
- Transanal endosurgical modified anorectal myomectomy is an effective, minimally invasive treatment option for selected cases of hypoganglionosis.
- A multidisciplinary approach is essential when evaluating children with recurrent infections and coexisting gastrointestinal symptoms.

Conclusion

Hypoganglionosis is a rare intestinal neuronal disorder typically presenting with chronic constipation, abdominal distension, or pseudo-obstruction. Extra-intestinal manifestations are uncommon and may obscure the underlying diagnosis. This case highlights the importance of considering occult gastrointestinal motility disorders in children with unexplained recurrent respiratory infections.This case highlights the importance of considering occult gastrointestinal motility disorders in children with unexplained recurrent respiratory infections. This case demonstrates how an uncommon gastrointestinal motility disorder can present with deceptively extra-intestinal symptoms, leading to diagnostic delay and significant morbidity. In this child, recurrent respiratory tract infections were traced to severe fecal retention caused by previously unrecognised hypoganglionosis. The case underscores the importance of considering occult gastrointestinal pathology when respiratory symptoms persist without a clear pulmonary explanation, particularly in the presence of chronic constipation or abdominal distension. Early recognition and targeted intervention are essential to prevent secondary complications. Transanal endosurgical modified anorectal myomectomy provided an effective, minimally invasive, and durable solution, resulting in complete resolution of both gastrointestinal and respiratory manifestations. This experience highlights the value of multidisciplinary assessment and reinforces the need for heightened clinical vigilance when evaluating children with recurrent infections and coexisting gastrointestinal symptoms.

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