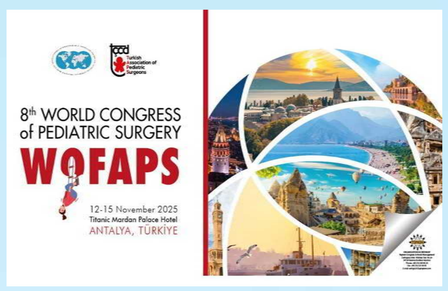


Late Diagnosis of Hirschsprung Disease: Causes and Outcomes

Berat Dilek Demirel, Pelin Nur Cinel, Başak Dağdemir Ezber, Sertaç Hancıoğlu, Beytullah Yağız

Ondokuz Mayıs University, Faculty of Medicine, Department of Pediatric Surgery, Samsun, Turkey

Aim: Hirschsprung's disease (HD) is a congenital disorder with typical presentation of delayed passage of meconium in newborns. Around 90% of cases are diagnosed within the first year. We aimed to evaluate the causes of late diagnosis and outcomes in patients diagnosed with HD ≥1 year of age.

Method: Patients diagnosed with HD ≥1 year of age between 2015 and 2024 were retrospectively evaluated for demographics, initial symptoms, causes of delayed diagnosis, surgical interventions, outcomes.

Results: Among the 92 patients operated for HD, 18 (19.56%) were diagnosed ≥1 year of age with a median age of 4.2 years (range: 1.42–14.17). M/F ratio was 13/5. In 72% (n=13) of cases, this was the first referral to pediatric surgery. Presenting symptoms were chronic constipation(n=18), infantile colic(n=3) and cow's milk protein allergy(n=3). One patient had osteogenesis imperfecta and one had Down syndrome. Only two patients (11%) had a history of delayed meconium passage. Three patients presented with enterocolitis, one with sigmoid volvulus. Abdominal distension was the most common physical finding. All patients underwent barium enema; three underwent anal manometry. Diagnosis was confirmed by full-thickness rectal biopsy. Short-segment HD was concluded in 12 patients, and ultrashort-segment in 6. Eight patients received colostomy. Three underwent transanal endorectal pull-through (TEPT), 3 laparoscopy-assisted TEPT, and rectal myectomy in 4 (2 needed TEPT later). Patients with colostomies underwent Soave-Boley pull-through after a median 4 months duration (range:2–10 months). During follow-up, 2 had enterocolitis, 3 required regular laxatives, 3 had fecal incontinence.

18 of 92 patients

≥1 year of age

19.56%

m/f:13/5

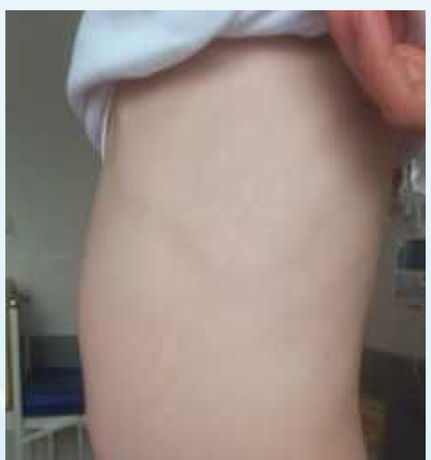
Median age 4.2 years

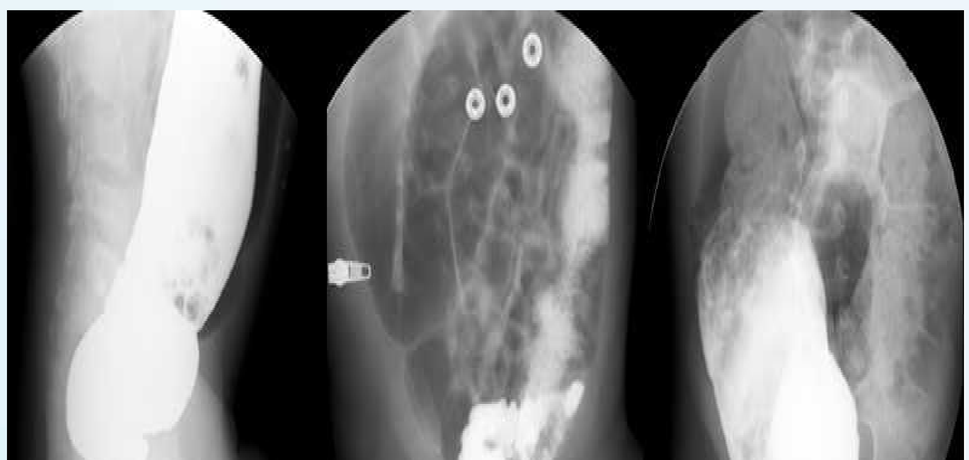
(1.42-14.17 years)

- **13 (72%) patients underwent initial pediatric surgical evaluation**
- **History of chronic constipation in all patients +**
- **Delayed meconium passage in 2 (11%) patients +**

3 patients with enterocolitis

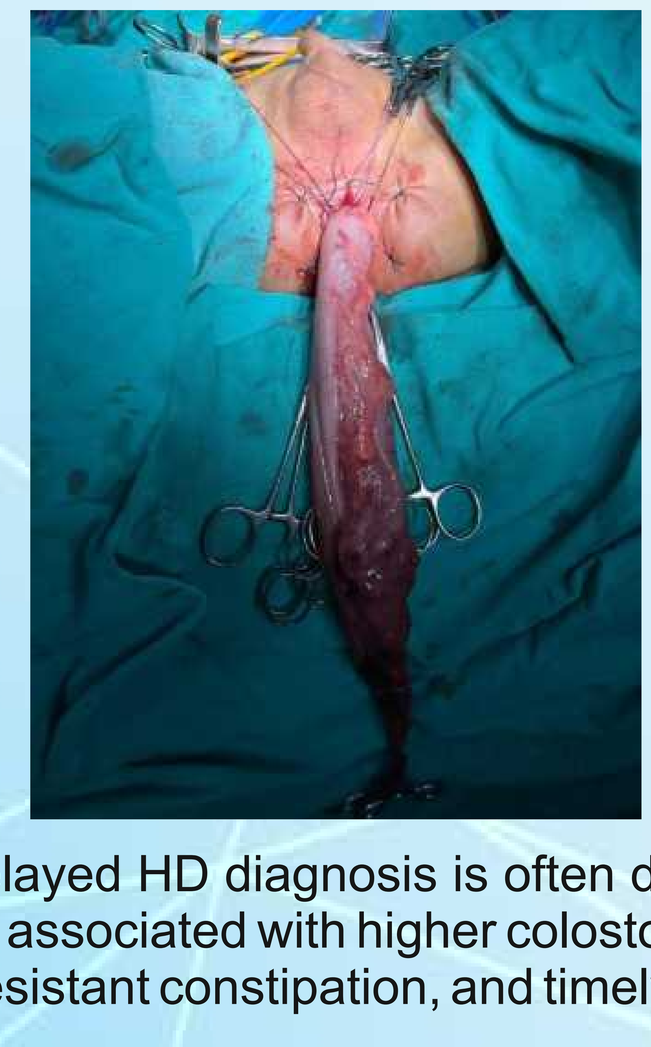
1 patient with sigmoid volvulus

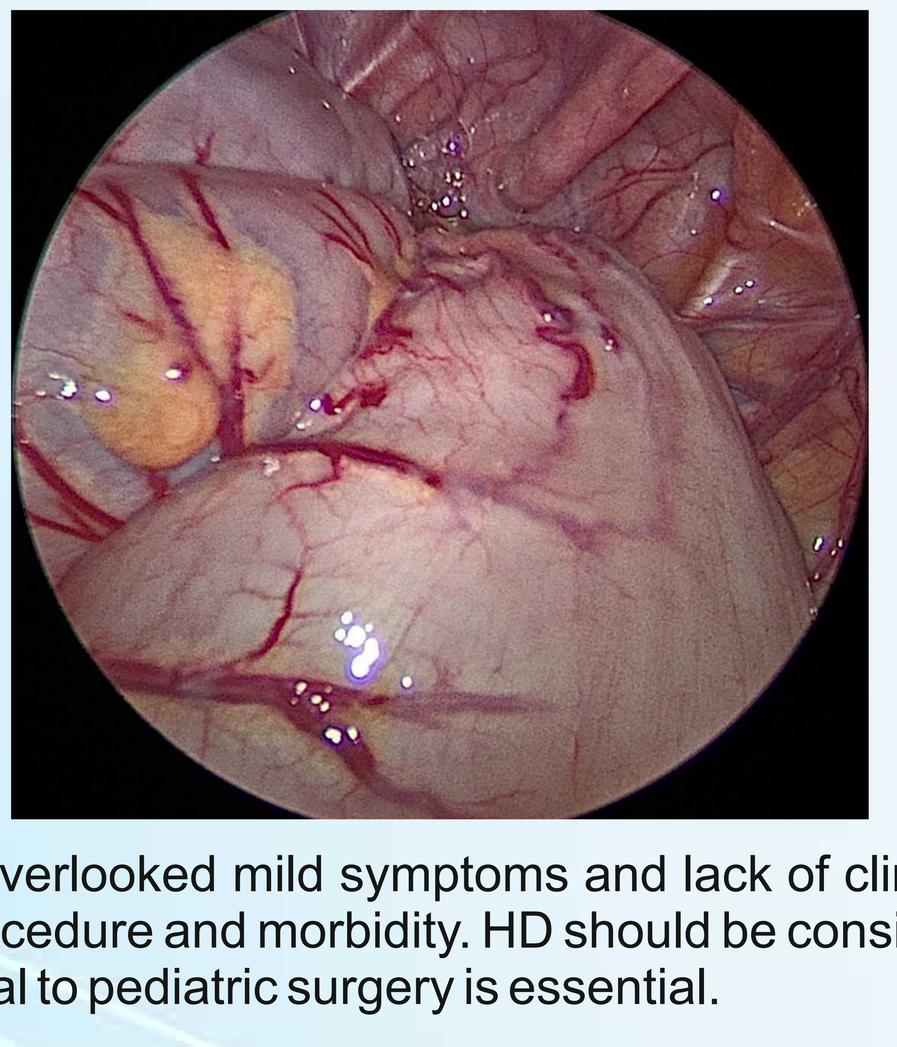




Short-Segment	12
Ultrashort-Segment	6
Colostomy	8
TEPT	3
Laparoscopy-assisted TEPT	3
Rectal Myectomy	4

Patient Characteristics and Procedures Performed
(Note: Two patients who underwent rectal myectomy subsequently required TEPT)





Conclusion: Delayed HD diagnosis is often due to overlooked mild symptoms and lack of clinical awareness. Late diagnosis is associated with higher colostomy procedure and morbidity. HD should be considered in patients with treatment-resistant constipation, and timely referral to pediatric surgery is essential.