## Case Report

# A Neglected Case of Hirschsprung's Disease Presenting in Adulthood : A Difficult Encounter

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Hirschsprung's Disease (HD) is characterized by the congenital absence of Ganglion Cells in all or part of the Colon. Neonate with HD presents with constipation since birth while chronic constipation is prominent in early childhood. The median age of presentation of the disease is 2-6 months. The condition is extremely rare in adulthood. We present an 18-year-old male who had features of long standing constipation and recurrent intestinal obstruction. We successfully treated it by diversion colostomy and rectal biopsy followed by modified Duhamel Operation. The rectal tissue biopsy report suggested HD. [*J Indian Med Assoc* 2021; **119(12):** 67-8]

***Key words*** **: Chronic constipation, Hirschsprung's disease, Adult, Contrast, Duhamel procedure.**

Hirschsprung's Disease (HD) is rare (1 in 5000 births) but, surgically correctable congenital disease. About 90% of the cases are diagnosed in the neonatal period or early infancy[1]. Sometimes, milder symptoms may go undiagnosed or misdiagnosed until adulthood and represented as HD in adulthood. Here, we present a case of HD in an 18-year-old male to highlight the diagnostic dilemma and difficulties in surgical management.

### Case Report

An 18-year-old patient presented with abdominal distention and chronic constipation since childhood with occasional acute exacerbation for which he was admitted to a rural hospital several times. He had a poor socio economic background with minimal family support. There was the expulsive release of liquid stool and air on performing a digital rectal examination. An abdominal x-ray revealed dilated gut loops up to the distal colon. Conservative management in the form of Nasogastric Aspiration, Saline Infusion and Enemas provided significant clinical improvement. Unfortunately, the patient left against medical advice before further investigations.

Three months later, he was re-admitted with similar symptoms. After re-suscitation, a contrast-enhanced Computed Tomography (CT) was done,which revealed a spectacular dilation of the proximal colon and a narrow terminal colon and rectum with a transition zone (Fig 1). In accordance with history, clinical findings and the CT report, we inferred it may be a case of HD presenting in adulthood. We took a consultation from our Paediatric surgical team and planned for two-stage surgical management.

***Editor's Comment :***

- The awareness of the possibility of HD in small children with constipation is important among doctors which reduces the delay in appropriate referral. The parents often may interpret it as a minor physiological deviation and remain falsely reassured.
- The parents must also be counselled regarding the benign nature of the disease and explained that surgery is the only available option for a possible cure with associated possible risks. The referral to the appropriate paediatric surgical team, suitable for the family, should be done without any delay.

Initially, we fashioned a leveling colostomy in the descending colon, excised the extraordinary large sigmoid colon up to a point just proximal to the rectum to create a distal Sigmoid Mucous Fistula. Full-thickness biopsy samples were taken from the rectum (spastic segment) and the colostomy site (descending colon) which showed a complete absence of intramural Ganglion cell and normal ganglion cells respectively. Postoperative (PO) recovery was uneventful and the patient was discharged on the 6$^{th}$ PO day.

The patient was re-admitted after 3 months for a modified Duhamel pull-through operation. The stoma was dismantled and the colon was brought down through retro-rectal space and an astonished at the level of 2 cm above the dentate line. We used two linear cutter gastrointestinal stapler (100 mm) to fashioned side-to-side anastomosis between the aganglionic rectal pouch and pull-through colon (posterior wall of the native rectal wall and anterior wall of the pull through the colon). The PO period was stormy as the patient developed additional medical complications due to Dengue viral infection. However, we managed the case successfully and could discharge the patient finally on the 28$^{th}$ PO day. At follow-up, after 2 months he was found to have developed a regular bowel habit with complete symptomatic relief, which was found to be maintained at 9 months follow-up too. Keeping in mind the patient's background and special needs, he is being closely followed up to avoid further neglect of any delayed complications.

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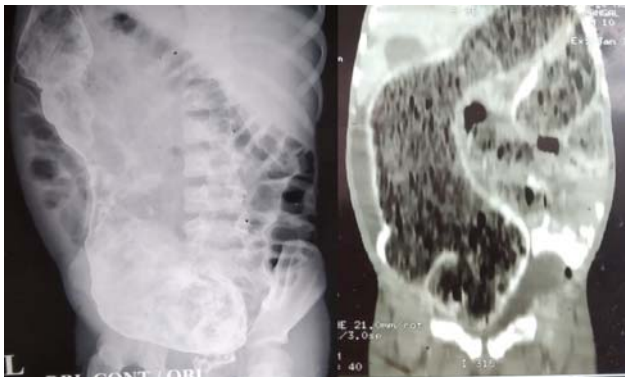

Fig 1 — Computed tomography with oral contrast showed hugely distended colon with distal narrowin

## Discussion

Diagnosis of the HD after 10 years was regarded as adult HD though, Natsikas *et al* made it 18 years[2]. In 1950, Rosin first described a case of HD in adult patients[3]. Grover *et al* stated that about 2% of patients with mild symptoms may go undiagnosed until adulthood. Acquired aganglionosis in adults due to neuronal degeneration by autoimmune response following parasitic infections can present with similar symptoms[4].

The symptoms of adult HD are similar to that of classic HD in childhood like; long-standing constipation and sometimes hospital admission for acute exacerbation. Additional history of dietary modification and psychosocial inhibition due to fecal soiling may be obtained[5]. Radiological evidence of distal narrowing and proximal dilatation in contrast (barium) Enema study (CES) is suggestive. However, CES sometimes become technically difficult in the adult because of the hugely loaded colon and or faecaloma. In such condition, a contrast CT is advocated[6]. The absence of recto-anal inhibitory reflex in anorectal manometry is another supportive evidence of HD. However, a rectal biopsy is a gold standard for the diagnosis of HD[7].

Management of adult HD practically a challenging task for General Surgeons because they are unfamiliar with the procedure as the surgery is hardly ever performed on adults. On the other hand, unfamiliarity with the adult organ proportions with a hugely dilated colon, posed a major challenge to the Paediatric surgeons too. A multidisciplinary approach (a collaboration of General and Paediatric Surgeons) is preferred to overcome these technical difficulties. Two-stage surgery is recommended to overcome the problems due to over-distended and elongated proximal colon[8]. Initial operation includes a leveling colostomy through the normally innervated colon, de-functioning stoma in the distal colon and rectal biopsy for tissue diagnosis[9]. The distal stoma permits cleansing of the caudal colon while the proximal colon reverts to near normal caliber, usually within 2 to 6 months (Fig 2). For definite reconstruction, Swenson abdominal pull-through,Soave endorectal pull-through and Duhamel pull-through procedures are described in the literature. Duhamel procedure is preferable when a considerable

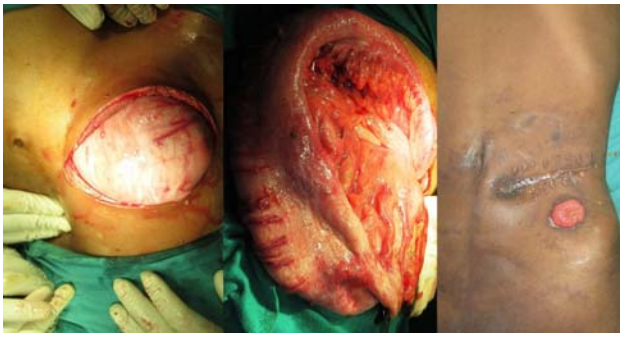

Fig 2 — Pictures during levelling colostomy and after three months showed hugely distended sigmoid

luminal discrepancy remains between the ganglionic and aganglionic segments of the colon[10]. Qiu *et al* showed that one-stage modified Martin-Duhamel or Rehbein's procedure may be a feasible surgical option for adult HD[11]. Irrespective of the surgical techniques, Postoperative bowel functioning is not always satisfactory. Patient's awareness about the complications (enterocolitis, constipation and fecal incontinence), regular check-ups in follow-up clinics and timely diagnosis of any unwanted complications are imperative to achieve good functional outcomes after surgery in adult patients.

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