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# HIRSCHSPRUNG'S DISEASE IN A SET OF MONOZYGOTIC TWINS *

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Aganglionic megacolon is a commoner cause of morbidity and mortality in infants than is generally realized. The symptoms in early life are bizarre and the diagnosis can be made early by a high index of suspicion in the presence of certain symptom complexes. Roentgen ray examination and rectal biopsy are the diagnostic aids in confirming such a diagnosis. After a definite diagnosis has been established, colostomy is mandatory until the child is old enough for a definite operative procedure. Delay of colostomy and use of temporizing measures may cause an unnecessary and unwarranted mortality.

The clinical picture of congenital megacolon is rather bizarre under the age of 6 months (table 1).[1] In the first week of life infants come to the hospital with a picture of intestinal obstruction, abdominal distention, vomiting and a history of passing little or no meconium. These are the classical signs and symptoms. Rectal examination reveals a tight sphincter and immediately after the examination, meconium or gaseous stools are emitted in an explosive manner. An upright film of the abdomen shows a distended bowel pattern with gas but there is no visible differentiation between the small bowel and the colon. In some cases fluid levels are seen with

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TABLE 1

PRESENTING SYMPTOMATOLOGY OF HIRSCHSPRUNG'S DISEASE UNDER SIX MONTHS OF AGE

| |
|---|
| GROUP I : BIRTH TO 6 DAYS<br>Intestinal obstruction |
| GROUP II : ONE WEEK TO 3 MONTHS<br>1. Septicemia<br>2. Peritonitis<br>3. Ulcerative colitis<br>4. Intermittent obstruction |
| GROUP III : THREE MONTHS TO 6 MONTHS<br>Malabsorption syndrome |

the increased distention. A barium enema aids in the diagnosis if the typical narrow segment in the rectosigmoid area, with an enlarged and distended proximal segment, exists. Sometimes this typical roentgen ray picture may not be seen because of the patient's early age but delay of evacuation of the barium from the bowel is characteristic of aganglionic megacolon, and it takes 24 to 48 hours for the evacuation of the barium.[1,3]

The patients in Group II (1 week to 3 months) show symptoms related to decompensation caused by the narrow colonic segment. A history of abdominal distention, obstipation and vomiting in the first week of life is almost always present. Later, diarrhea develops; patients become dehydrated and feverish, usually cyanosis is evident. The development of septicemia, peritonitis, ulcerative colitis and intermittent obstruction causes death unless treated early and properly.

Patients over 3 months old manifest classical signs and symptoms with abdominal distention, emesis and constipation accompanied by episodes of diarrhea. These cases are usually suspected of malabsorption syndromes such as celiac disease and feeding problems. A rectal examination reveals an empty rectal segment followed by an explosive foul stool when the examining finger is removed. The diagnosis may be confirmed by a barium enema and a rectal biopsy.

The differential diagnosis under the age of 6 months is summarized in table 2. The complications [4] are shown in table 3.

In this paper we will present a set of monozygotic twins born at term after an uncomplicated pregnancy and delivery. This was the first pregnancy of a thirty year old mother. There was a single placenta with two umbilical cords. Within a few days both babies presented symptoms of an acute intestinal obstruction.

## *Case Reports*

**Case I.** U. E., a four day old boy, was admitted with a chief complaint of emesis. His birth weight was 2350 Gm. and he had started breast feeding after 24 hours of life. On the first day he passed very little meconium. The emesis started when he was three days old; it was non-projectile, repeated every ten minutes, and contained green matter. He did not pass any stool until he was four days old and his abdomen became distended on the day of admission to the hospital.

**Physical examination:** upon admission the baby's weight was 2020 Gm., height 46 cm., temperature 37.8 C. rectally, respiration 36 per minute and pulse 140 per minute. His turgor had decreased considerably, his tonus was normal and both sucking and Moro's reflex were good. His abdomen was slightly distended but the umbilical cord was on and clean. The bowel sounds had decreased and during the examination he vomited some green material.

A diagnosis of intestinal obstruction, possibly ileal atresia, was made, however, congenital megacolon was not ruled out.

All laboratory results were within normal limits. See table 4.

### TABLE 2

DIFFERENTIAL DIAGNOSIS OF AGANGLIONIC MEGACOLON

| |
|---|
| A. BIRTH TO FIRST WEEK |
| 1. Acute intestinal obstruction |
| a) Duodenal atresia |
| b) Jejuno-ileal atresia |
| c) Malrotation |
| d) Meconium ileus |
| 2. Lesions of rectum and anus |
| a) Anal stenosis or stricture |
| b) Valve-like lesions in rectum and sigmoid |
| B. AFTER FIRST WEEK |
| 1. Feeding problems |
| 2. Constipation |
| 3. Infections |
| 4. Malabsorption syndrome (celiac, etc.) |

TABLE 3

COMPLICATIONS OF AGANGLIONIC MEGACOLON

| |
|---|
| A. PREOPERATIVE |
| 1. Enterocolitis and dehydration |
| 2. Septicemia |
| 3. Generalized peritonitis |
| B. POSTOPERATIVE |
| 1. Peritonitis due to leaks at the suture line |
| 2. Ileus |
| 3. Wound infection |
| 4. Adrenal insufficiency |
| 5. Septicemia |
| 6. Enterocolitis |
| 7. Stricture formation |
| 8. Pelvic abscess |
| 9. Others |

**Radiology :** An upright flat film of the abdomen showed some gas and fluid levels with a distended bowel pattern. Figure 1 a.

The upper gastro - intestinal examination, using barium, disclosed that radiopaque material was regurgitated into the esophagus after passing to the stomach. Figure 1 b.

**Figure 1 a.**

**Figure 1 b.**

The patient was discharged at his parents' request on the day after admission. He had been given whole milk while in the hospital and passed 5 stools, 4 of which were watery. He had also vomited 4 times. He was given penicillin and streptomycin intramuscularly.

TABLE 4

CASE 1

| NAME | AGE | WEIGHT | CHIEF COMPLAINT | HISTORY OF PRESENT ILLNESS | PHYSICAL FINDINGS | LABORATORY FINDINGS | ROENTGEN RAY FINDINGS | THERAPY |
|---|---|---|---|---|---|---|---|---|
| U.E. No. 1 Boy | 4 days | 2020 Gm. | Vomiting, Abdominal distention. | Greenish emesis, 1 day. Passed small amount of meconium in first day. No more meconium passed until 4 days old. | T. 37.8 C. Abdomen distended, bowel sounds decreased. | Hemoglobin : 25 Gm. per 100 ml. RBC : 5 million WBC : 8000 Differential : lymph. 42 % neutro. 58 % | Flat film of abdomen-upright: gas, fluid levels. Upper GI : Regurgitation of barium into the esophagus. | Antibiotics. |
| U.E. No. 1 (2nd admission) | 10 days | 2000 Gm. | Inability to pass stools. Abdominal distention. Cyanosis. | Did not have any bowel movement for preceding 5 days. Abdominal distention increased. | T. 39 C. Dehydrated. Turgor decreased. Abdomen very distended. Venous pattern present. Loops seen over the abdominal wall Bowel sounds present. Omphalitis. | Hemoglobin: 16.25 Gm. per 100 ml. RBC: 4.8 million WBC : 22,000 Differential : neutro. 52 % lymph. 48 % Urine normal. Blood group: O, Rh positive. | Barium given 5 days before still present in the colon. Colon narrow up to splenic flexure. | I. V. fluids. Antibiotics. Blood transfusion. Oxygen. Gastric decompression. Transverse colostomy. |

TABLE 5

CASE 2

| NAME | AGE | WEIGHT | CHIEF COMPLAINT | HISTORY OF PRESENT ILLNESS | PHYSICAL FINDINGS | LABORATORY FINDINGS | ROENTGEN RAY FINDINGS | THERAPY |
|---|---|---|---|---|---|---|---|---|
| U.E. No. 2 Boy | 10 days | 2060 Gm. | Vomiting 2 days. Abdominal distention. Diarrhea 3 days. Fever. Cyanosis. | Did not pass stools until 4 days old then passed very little meconium. At age of 8 days fecaloid emesis started and diarrhea developed. Stools green, watery, 8-9 per day. | T. 39 C. Cyanotic. Abdomen distended and bowel-loops seen over the abdominal wall Bowel sounds absent. Rectal examination: No tightness. Passed stool after examination. | Hemoglobin 14.45 Gm. per 100 ml. RBC : 5 million WBC : 10,000 Differential : neutro. 47 % lymph. 53 % Serum Co2 23.2 mEq/L. Serum Cl 100.8 mEq/L. Blood group : O, Rh positive. Urine : normal. | Flat film of the abdomen, upright: Gas-fluid levels. No gas seen in rectum. Possibly small intestines distended. | I. V. fluids. Blood transfusions. Oxygen. Antibiotics. |

Six days after discharge from the hospital (table 4) he was readmitted because of excessive abdominal distention, cyanosis and inability to pass stools, during the entire period after discharge.

**Physical examination :** Upon readmission the baby's temperature was 39 C. pulse 120 per minute, respiration 30 per minute; his weight was 2000 Gm., Moro's reflex and sucking were positive, turgor had decreased and tonus was normal. Both pharynx and tonsils were hyperemic. His abdomen was extremely distended with venous pattern. Distended loops of the bowel were visible over the abdominal wall and there was omphalitis.

The diagnosis was aganglionic megacolon, omphalitis, possible sepsis and dehydration.

There were no remarkable laboratory findings.

**Radiology :** A flat film of the abdomen (fig. 1 c) disclosed the presence of radiopaque material, given 5 days before, in the cecum, transverse colon, ascending and descending colon. The bowel loops before the splenic flexure looked enlarged and that part of the bowel between the rectum and the splenic flexure seemed to be narrow, (fig. 1 d). Under fluoroscopy a barium enema was attempted but was not successful because of the return of the barium from the rectum.

With these clinical and roentgen ray findings a diagnosis of Hirschsprung's disease was made and the baby was operated on under 1 per cent procaine local anesthesia, because of his poor condition. During surgery the transverse colon was found to be very much enlarged up to the splenic flexure and the

**Figure 1 c.**

**Figure 1 d.**

colon beyond this point was narrow. A transverse colostomy was made. The baby had an apneic spell at the end of the operation. This was treated with intubation and oxygen successfully.

A small blood transfusion, intravenous fluids, penicillin and streptomycin were administered postoperatively and the patient was kept in oxygen in an isolette.

During the first postoperative day, the colostomy seemed to be working well. In addition to the intravenous fluids, a small amount of whole milk was taken by mouth successfully. A small blood transfusion was again given. On the third postoperative day the patient tolerated oral feeding well and the intravenous fluids were discontinued. Formed stools, 5 to 7 times daily were passing through the colostomy opening. On the fourth postoperative day, the patient had a sudden apneic spell and expired in spite of artifical respiration, oxygen and adrenaline. The postmortem examination revealed diffuse atelectasis in both lungs. The serosa of the intestines was hyperemic and edema-

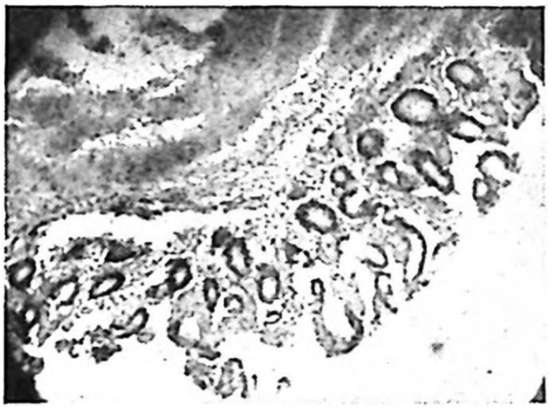

**Figure 2.**

tous. The transverse colon was enlarged to the splenic flexure; the descending and sigmoid colons and the rectum appeared narrow. There was local peritonitis around the colostomy area in the transverse colon.

The microscopic examination showed enterocolitis (fig. 2) in the ileum, jejunum and transverse colon. The mucosa contained scattered areas of bleeding. No ganglion cells were seen (fig. 3) between the circular and longitudinal

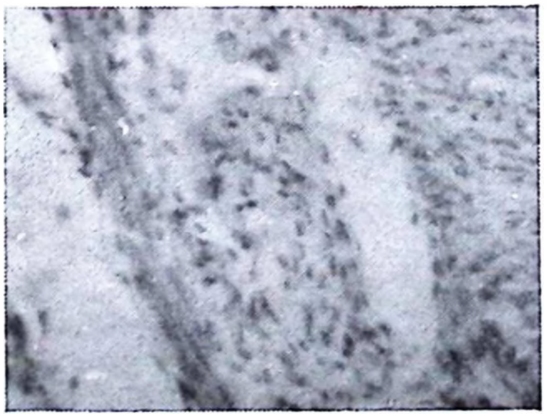

**Figure 3.**

layers in the multiple sections made from the rectum, sigmoid and descending colon. Extensive hemorrhage was visible in both adrenal cortices.

**Case 2.** U. E. II, brother of case 1, was admitted at 10 days of age with complaints of abdominal distention, emesis, diarrhea, fever and cyanosis. He

had passed no meconium until he was 4 days old at which time only a small amount was passed. He had been started on breast feeding when he was 24 hours old. Emesis which was fecaloid in character had increased to 3 or 4 times daily by the time he was 8 days old. Two days prior to his admission to the hospital he began to have diarrhea with watery stools 8-9 times daily. On the day of admission his abdomen became distended, his temperature rose and his color was blue.

**Physical examination:** Though his birth weight was 2750 Gm., on admission it was 2060 Gm., his pulse was 160 per minute and respiration 40 per minute. His color was cyanotic, turgor was greatly decreased, Moro's reflex was absent and sucking was present but very weak. Fine rales were heard over the lung fields, his abdomen was greatly distended with loops of the bowel clearly visible over the abdominal wall. Bowel sounds were absent. The rectal examination disclosed no tightness and a stool was emitted immediately after withdrawal of the examining finger.

Acute intestinal obstruction, possible ileal atresia, bronchopneumonia, dehydration and possible sepsis were diagnosed.

All laboratory findings were within normal limits.

**Radiology:** An upright flat roentgenogram of the abdomen showed many gas and fluid levels and distended intestinal loops. No gas was seen in the rectum, (fig. 4).

The baby was started on intravenous fluids, penicillin and streptomycin and placed in an incubator in oxygen. A small blood transfusion was given but general condition remained poor. He died 6 hours after admission.

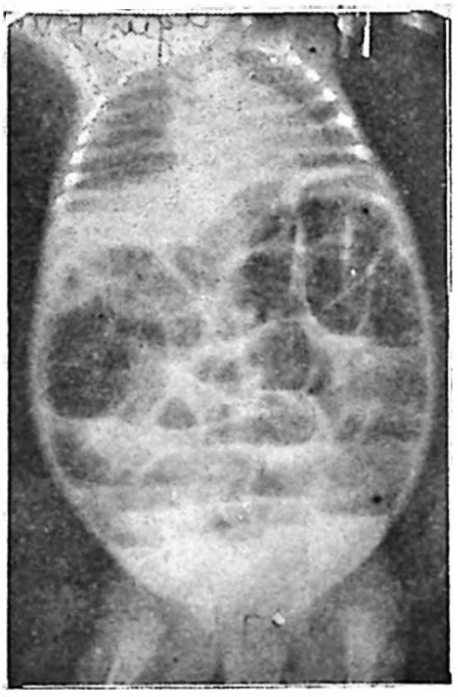

**Figure 4.**

The postmortem examination showed that the peritoneal cavity contained some bloody fluid and the serosa of the intestines was hyperemic and shiny. The cecum was enlarged but the ascending transverse colon appeared to be of

normal size. The descending colon seemed to grow narrow from above to below and the diameter of the lowest point was 1 cm. An area of bleeding around the rectum about 2 × 3 cm. was found. Other organs and the ureters were normal.

Examination under the microscope revealed that the transverse colon and the jejunum exhibited a slight edema and an inflammatory infiltration in their submucosa and serosa. Ganglion cells were present between the transverse and longitudinal muscle layers. The mucosa of the rectum showed partial bleeding. There was a thickening of the submucosa, enlargement of the vessels and a heavy inflammatory infiltration. The sigmoid colon had similar characteristics. There were no ganglion cells in the multiple sections of the rectosigmoid area reviewed.

The spleen and kidneys showed passive congestion. The adrenals exhibited lipoid depletion and the liver was fatty with passive congestion.

## *Discussion*

It is claimed that Hirschprung's disease is familial [5,6] and the chances of having aganglionic megacolon is much higher in the male siblings of a patient than in the female. The statistical analysis done by Bodian and colleagues showed that cases of the disorder occurring in subsequent female children are few and in subsequent male children about 1 in 5.[5] To our knowledge, few reports have appeared in medical literature concerning the involvement of this pathology in twins. (Table 6).

In 1920, Popper [7] reported on a pair of female twins. One had passed no meconium and died at 4 days from intestinal obstruction. There was no necropsy. The sister twin was treated with repeated enemas for retention of meconium and was reported alive at 28 weeks though with a distended abdomen and visible peristalsis. The barium enema film showed a typical narrow segment of average length. No evidence is given as to whether these twins were monozygous or dizygous. It was considered that the first twin probably had Hirschpsrung's disease and the roentgen ray evidence on the second twin proved the diagnosis.

In 1951, Bodian and his colleagues [5] published a report of aganglionic megacolon in two sets of twins. The first set consisted of 40 month old brothers. One twin was well until he was 1 year old, then he became constipated to the extent that daily laxatives were required. His weight gain was normal but his abdomen was slightly more enlarged than that of his twin brother and he was slightly taller. A barium enema revealed a typical short narrow segment affecting the rectum.

## TABLE 6
## REPORTED CASES OF AGANGLIONIC MEGACOLON IN TWINS

| | |
|---|---|
| J. POPPER (1920) | |
| A - girl | Passed no meconium and died at 4 days from intestinal obstructions. No necropsy done. |
| B - girl | Treated with repeated enemas for retention of meconium. She was reported alive at 28 weeks with a distended abdomen and visible peristalsis. Ba enema: a typical narrow segment of average length. (It is not reported whether the twins were monozygous or dizygous.) |
| M. BODIAN ET AL. (1951) | |
| A - boy | 3 ½ years old. Constipation, abdominal distention started at 1 year. Weight gain was normal. |
| | Ba enema: a typical short narrow segment affecting rectum. |
| B - boy | 3 ½ years old. Emesis after each feeding started at 7 weeks. Admitted to a hospital for 6 weeks. Constipation began at 1 year and necessitated laxatives. Abdomen became protuberant after age of one year, weight gain was normal. |
| | Ba enema: a short narrow segment affecting the rectum. Their 6 year old brother had Hirschsprung's disease. |
| A - boy | 12 years old. Constipation and abdominal distention. Smaller than his brother. |
| | Ba enema: A short narrow segment near the rectum. |
| B - boy | Well, without any complaints. |
| | They probably are dizygous. |
| PRESENT REPORT (1961) | |
| A - boy | 10 days old. Abdominal distention. Passed little meconium. Roentgen rays: gas-fluid levels in flat film. Delay of evacuation of barium. Pathology: no ganglion cells in myenteric plexus in rectum, sigmoid and descending colons. |
| B - boy | 10 days old. Emesis, abdominal distention, diarrhea. Did not pass meconium until 4 days old. |
| | Roentgen rays: gas-fluid levels in flat film. No gas in rectum. Pathology: ganglion cells absent in myenteric plexus in rectosigmoid area. |

The other twin of this set began to vomit at every feeding when he was 7 weeks old. He was hospitalized for 6 weeks, constipation began at 1 year and gradually became so obstinate as to require daily laxatives. His weight gain, also was normal. His abdomen was protuberant after his first year. He became pot-bellied with a tympanitic abdomen in which fecal masses were palpable. A barium enema revealed a short narrow segment affecting the rectum. A transverse colostomy was done on both twins. Their 6 year old brother also had Hirschsprung's disease.

The second set of twins mentioned by Bodian et al. consisted of 12 year old boys. One had constipation and abdominal distention and was smaller than his brother. A barium enema showed a short narrow segment near the rectum. His twin had no complaints. According to the studies done for identification, they were dizygous.

Our twins were both male and had one placenta with two cords. Their blood group was O, Rh positive, and both presented a picture of acute intestinal obstruction in their first days of life. Histologic examination revealed an absence of ganglion cells in the myenteric plexus involving the descending and sigmoid colons and the rectum in one brother and the rectosigmoid area of the second brother. No other member of the family had ever had Hirschsprung's disease.

*Summary*

Hirschsprung's disease in a set of newborn twins is presented and clinical symptomatology and diagnosis of congenital megacolon is briefly reviewed.

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