CASE REPORTS

GASTRIC DILATATION, MEGACOLON, AND IDIOCY IN IDENTICAL TWINS

BY

D. S. FAIRWEATHER, M.A., M.D., M.R.C.P., D.P.M., and
H. J. L. O'SULLIVAN, M.B., B.Ch., B.A.O.
Stoke Park Colony, Stapleton, Bristol

The chief interest in the cases to be described lies in the association of atonic dilatation of the stomach, megacolon (Hirschsprung's disease) and the coeliac syndrome with idiocy, and in its occurrence in identical twins.

Family and Social History

The parents were well educated, the father being a master at a well-known public school. There were no mental or physical abnormalities of significance, apart from a history of twinning among cousins of one of the parents.

The first child of the family was born with difficulty, labour being long and terminated by forceps, her subsequent development, both physical and mental being normal. The twin conception took place eight months after the birth of the first child. For four months the mother was able to carry out her household duties in a normal manner, but at the end of that time her abdomen enlarged markedly. She felt sickly and ill, with vague indigestion and abdominal discomfort, which necessitated long periods in bed. Labour commenced March 16, 1938, six to seven weeks before full term, and was characterized by one pain for each birth with thirteen minutes between them. An excess of liquor amnii was passed, followed by a single placenta with two umbilical cords attached. A third pregnancy terminated uneventfully two years later, a normal child resulting.

Early History

The birth weights of the twins were identical, being 5 lb. 12 oz. each. On the third day both of them developed asphyxia, and went blue several times, the younger baby causing considerable anxiety that lasted throughout the day. The condition never recurred. Neither child was able to take the breast, though they were fed on breast milk from the bottle up to the end of the sixth week, when they were put on Cow and Gate. They gained weight very slowly, and at the end of the fifth week the elder (A) weighed 6 lb. 9 oz. and the younger (B) 6 lb. 6 oz. From then onwards their weights

increased more quickly, so that in the sixteenth week they weighed 10 lb. 2 oz. and 10 lb. respectively.

Their bowel history up to this time gave no cause for alarm, but at five months both of them contracted what was called by the visiting practitioner 'summer diarrhoea.' In the case of B, this was followed by constipation with considerable abdominal discomfort that lasted twenty-four hours. Thereafter both of them became liable to alternate diarrhoea and constipation, occasioned by such trivial changes in the diet as alternating from Robinson's patent groats to Robinson's patent barley.

Both children were delicate and liable to coldness of the extremities. During teething, which began at ten months, B was liable to a rise of temperature up to 101° F., and at fifteen months A developed bronchitis and laryngitis with high fever. At seventeen months B began to crawl, but her sister made no efforts to do so. The delay in development was attributed to prematurity at birth, so that the possibility of mental deficiency was not entertained until they were about eighteen months old. Treatment with thyroid extract was instituted at this time, but no benefit resulted apart from some improvement in the condition of the peripheral circulation.

At twenty-one months they were seen by Sir Leonard Parsons, who diagnosed mental deficiency which he attributed to primary amentia. He reported that he could find no other cause for their backwardness, and that at this time they presented no indications of chronic abdominal disease. In their third year, however, the parents noticed that their abdomens at times were distended.

At two years five months the anterior fontanelle was not quite closed in the case of the elder twin, and had only just closed in the case of the younger. It was noted at this time that they showed a lack of initiative and imitiveness, that they appeared indifferent to the objects around them, and that they were unable to recognize their mother. They made no attempt at walking, talking, or feeding themselves, and their habits with regard to urination and defaecation were undeveloped.

At two years nine months B began to show some interest in walking, and before her admission to the

TABLE 1

	A	B
General appearance	In poor health, though of good colour and fairly well nourished	In poor health, though of good colour; rather thin
Height	33 in.	34½ in.
Weight	21 lb.	21 lb.
Eyes	Blue	Blue
Tongue	White fur	Clean
Teeth	Healthy	Healthy
Throat	Tonsils enlarged	Tonsils enlarged
Thyroid	Not enlarged	Not enlarged
Skin	Scarring on buttocks and vulva	Healthy
Heart	Apex beat in 5th interspace and mid-clavicular line. No enlargement of area of cardiac dullness. Tic tac rhythm. No adventitious sounds. Poor tone	Apex beat in 5th interspace. No enlargement of heart. Tic tac rhythm. Rapid action. No adventitious sounds
Lungs and respiratory system	Fair respiratory range. Slightly impaired percussion note. Harsh breath sounds; no adventitious sounds. Slight cough, and nasal discharge	Good respiratory range. No impairment of percussion note. Harsh breath sounds
Abdomen	Poor musculature of abdominal wall. Liver, spleen, and kidneys not palpable. Distension very marked. Flatus +. Small umbilical hernia. Considerable evidence of constipation	Divergence of recti. Thin musculature of abdominal wall. Liver, spleen, and kidneys not palpable. Distension very marked. Flatus ++. Small umbilical hernia
Lymphatics	Small discrete glands palpable in both groins, left axilla, and left cervical region	Small discrete glands palpable in both groins, axillae, and cervical regions
Optic discs	Impossible to examine	Impossible to examine
Pupils	Equal, regular, and central. React normally to light	Equal, regular, and central. React normally to light
Motor functions	Sits up and stands with support. Is unable to roll over on to her abdomen. Is unable to pick up a cube from the table or to hold two cubes. Shows no preference for right or left hand. Does not creep when laid prone. Is unable to walk, but makes stepping movements. Rocks from side to side continuously when in bed. Some purposeless movements of hands. Myoclonus present	Sits up and stands with support. Can roll over on to her abdomen. Is unable to pick up a cube from the table, or to hold two cubes. Shows no preference for right or left hand. Creeps when laid prone. Walks with difficulty, is slightly spastic, and has poor balance. Lies in bed in the opisthotonos position and swings herself, balanced on head and feet. Myoclonus present
Reflexes	Abdominal reflexes: present but diminished. Knee jerks: present and equal. Plantars: flexor (R. and L.)	Abdominal reflexes: not elicited. Knee jerks: present and equal. Plantars: flexor (R. and L.)
Sphincters	Wet and dirty in habits	Wet and dirty in habits
Sensory and receptive functions	No evidence of gross dysfunction of sight, hearing, or crude sensation, but does not understand anything that is said to her	No evidence of gross dysfunction of sight, hearing, or crude sensation, but does not understand what is said to her
Urine	No abnormal constituents	No abnormal constituents
Wassermann reaction	Negative	Negative

Colony she could walk with very little help, though she still showed no interest in toys of any description. A, however, remained greatly retarded in this respect, and in general appears to have been rather more backward and lethargic than her sister. She could balance herself sitting up, but could not pull herself up to the sitting position and was liable to throw herself backwards. Her chief occupation was watching her hands, which she used aimlessly though she soon tired.

At three, both of them developed whooping cough.

Condition on Admission

They were admitted to Stoke Park Colony on June 11, 1941, at the age of three years and three months; their physical and mental states were as given in table 1.

Attempts at mental testing established the incapacity of the twins for any degree of purposive activity. They were both unable to talk, or do anything for themselves. A refused to lie down. She would wave her hands aimlessly in the air, took no notice of words addressed to her, and would fall asleep swaying her body. She was miserable and fretful, and when offered toys she pushed them all away and refused to take anything into her hand. Both were quite incapable of doing any of the Terman-Merrill or Merrill-Palmer tests. B played with bricks only by throwing them on to the floor. She was very restless, and usually wanted to move

about touching things all the time. On some occasions she spent much time gazing at one hand, but at other times she took no interest in her surroundings, was blank, salivated excessively, and cried frequently. They were both diagnosed as idiots, and were incapable of guarding themselves against common physical dangers, or of being taught to do so; their mental ages were assessed as considerably less than two years.

Progress after Admission

The twins were very liable to sickness, at first after almost every feed. The vomitus was offensive and mixed with sputum. A vomited even after small quantities of food.

There was some alternation in the frequency of their motions, towards diarrhoea and constipation, with corresponding alternation in their consistency, between fluidity and more solid whitish putty-like stools, when they were very foetid, so that 'it was almost impossible to go near.'

Both continued to show marked abdominal distension, this being more marked in the case of B than of A, and in order to relieve it enemata and the passage of the flatus tube were necessary from time to time. Their progress was interrupted by respiratory infections and what were considered to be bouts of intestinal toxæmia with fever, restlessness, recurring distension, constipation, and fretfulness. Coccal infections of the skin, such as boils

TABLE 2

	A	B
Height	39 in.	39½ in.
Weight	29½ lb.	31½ lb.
Skin	Multiple scarring from coccal infections	Multiple scarring from coccal infections. Extensive bald patches on the scalp
Heart	Apex beat localized in 4th left intercostal space, just internal to the nipple line	Apex beat localized in 5th left intercostal space, just internal to the nipple line
Lungs	No abnormal signs except for tympanitic resonance as high as the 8th left intercostal space behind, presumably due to gastric dilatation	No abnormal signs
Abdomen	Abdominal circumference variable in extent, average about 25½ in. Umbilical eversion. Distended abdominal veins. Gross abdominal distension. Abdominal palpation followed by considerable expulsion of foul-smelling flatus	Abdominal circumference variable in extent, average about 23 in. Abdominal distension less marked than in A
Central nervous system	Facial movements normal except for occasional twitching of right side. Manneristic rocking of head from side to side. Hyporeflexia affecting arm and ankle jerks. Positive Hoffman's sign—left. Walks with help with feet everted and bilateral pes planus. Urinary and faecal incontinence	Hippus occasionally noticed. Assumes opisthotonos position lying in bed. Absent triceps jerks and abdominal reflexes. Positive Hoffman's sign right and left. Normal gait. Urinary and faecal incontinence less marked than in A

TABLE 3

	A	B
Blood counts	Red cells 5,010,000 Haemoglobin 80% Colour index 0.80% White cells 10,300 Polymorphs 32% Lymphocytes 65% Large mononuclears 3% Eosinophils —	4,700,000 85% 0.91% 8,400 50% 42% 4% 4%
Sugar tolerance	Fasting 71 mg. % ½ hour 228 mg. % 1 hour 284 mg. % 1½ hours 316 mg. % 2 hours 198 mg. %	79 mg. % 238 mg. % 292 mg. % 224 mg. % 174 mg. %
Dose : 50 g. Dextrose		
Blood calcium	9.5 mg. %	10.8 mg. %
Faeces (48-hours' excretion)	A fatty clay-coloured stool (weight 83 g.) of uniform consistency cutting like a fairly firm slab of butter. Dry matter 38.5%. Microscopic examinations revealed excess of soap crystals and plaques. No starch granules detected. Few partially digested meat fibres and cellular residues. Stercobilin much decreased (about ½ normal). No bilirubin. Total fat: 30.4% of dried faeces. (Soap 20.8%. Fatty acids 7.6%. Neutral fat 2%.) Culture: <i>b. coli</i> and <i>b. faecalis alkaligenes</i> . No tubercle bacilli.	A fatty clay-coloured stool (weight 71 g.) of uniform firm consistency. Dry matter 33.8%. Microscopic examination revealed excess soap crystals and plaques. No starch granules detected. Few partially digested meat fibres and cellular residues. Stercobilin much reduced (about ½ normal). No bilirubin. Total fat: 30.6% of dried faeces. (Soap 23.8%. Fatty acids 4.6%. Neutral fat 2.2%.) Culture: <i>b. coli</i> and <i>b. faecalis alkaligenes</i> . No tubercle bacilli.

made their appearance from time to time, and B in particular was very liable to pull out her own hair from the scalp.

Their weights showed considerable variation with a tendency to alternate increases and decreases. During their six years in the Colony, A has gained 8½ lb. and B 10½ lb., in contrast with the normal average increment during that period of 28 lb.

(For photographs see Plate Va and b).

The present condition of the twins is given in table 2, and the laboratory findings in table 3.

Radiological findings of A. Radiography was complicated by the fact that the patient was a resistive idiot, but it was quite apparent that there was a tremendous ballooning of the stomach which led to upward displacement of the heart shadow (Plate Vc). The bones showed definite evidence of osteoporosis, especially in the long bones.

She was reluctant to take the whole quantity of barium meal and took two and a half hours to take 12 oz. A film at the end of two hours showed a forward rotation of the stomach which subsequent radiographs confirmed (Plate Vd). As the child had to be radiographed in a recumbent posture it seemed probable that the x-ray appearances were distorted by the projection of the spinal column against the stomach, this giving the impression of two separate loculi (Plate Ve). In this film at the end of four hours the greatly distended outline of the stomach was apparent with a small quantity of barium in the

left 'loculus,' and a larger quantity in the right 'loculus.' Barium is seen in the small intestine, in which there is no evidence of dilatation. A film (Plate Vf) taken at the end of five and a half hours, showed the left 'loculus' to be empty, with the right 'loculus' partly emptying through the pyloric outlet, which appears to be situated more internally and posteriorly than usual. The colon began to fill at the end of five hours, and at the end of twenty-four hours most of the barium was situated in the ascending and pelvic colons.

A barium enema showed marked redundancy of the descending colon as well as considerable ballooning. The x-ray appearances, therefore, may be summarized as follows: There is marked displacement of the heart shadow upwards, due to enormous dilatation of the stomach and ballooning of the colon. There is evidence of redundancy of the colon as well as of megacolon (Plate Vg).

Radiological findings of B. Radiographs of the chest showed some upward displacement of the heart but not to the extent of that in A. The same tendency to biloculation was noted, but the emptying time of the stomach was much less than in A's, and half an hour after the barium was given there was a considerable amount of it in the coils of the small intestine (Plate Vlh). The same effect produced by gastric dilatation was found with the duodenal bulb pointing internally and backwards (Plate VIj).

At the end of four hours the stomach was almost empty and at the end of four and a half hours the ascending colon was beginning to fill and was evidently dilated. At the end of twenty-four hours the meal was almost solely confined to the pelvic colon and rectum.

A radiograph after enema of 30 oz. of barium mixture showed marked reduplication of colonic shadow to the right, which was dilated in the whole of its extent up to the splenic flexure (Plate VIk).

Mental progress. Their scores by the Gesell standards best illustrated the mental progress of the twins while in the Colony, and these are summarized in table 4.

Evidence of uniovularity. In support of the twins being identical we have the mother's statement, confirmed by the doctor in attendance, of a single placenta. As regards other characteristics, we may summarize them as follows:

APPEARANCE AND GENERAL FEATURES. There is no doubt, as indicated by their photographs, that their general similarities of physique have been considerably modified by the various vicissitudes of their physical condition. The older twin presents a more angular facial outline with slight facial tic, while the younger has more rounded features.

HAIR COLOUR, TEXTURE, AND FORM. Their hair colour was identical. Two distinct shades were apparent, corresponding on Fischer's Scale to F11 (dominant) and F8 (subordinate). A's hair was of slightly finer texture than B's, which was attributed to her poorer nutrition. The hair of both was curly.

EYE COLOUR AND IRIS PATTERN. Both had the same shade of bluish irides without any marked features of iris pattern.

SKIN COLOUR AND COMPLEXION. Both had fresh complexions and skin of fine texture, though again some differences were attributable to the different state of their nutrition.

EYELASHES, EYEBROWS, LIPS, SHAPE OF EARS, SHAPE, SIZE, AND ARRANGEMENT OF TEETH. Eyelashes

and eyebrows were of an identical colour, form, and texture. The lips showed slight dissimilarities in that B's were rather thicker, and A's were slightly contorted by the facial tic, though their underlying structure was the same as B's. The shape of the ears was identical, though B's were slightly more fleshy. The shape and size of the teeth were identical but there were differences in arrangement which could have been due to variations in eruption consequent on faulty nutrition.

HANDS AND FINGERS. Shape and size showed close similarities. Attempts at procuring finger prints were not very successful and ridge counts were impossible to obtain. Finger nails and lunulae were identical.

MIRROR IMAGING. We could find no evidence of mirror imaging in regard to handedness, dental irregularities, and palm patterns, though evidence was found of mirror imaging in the pattern of the hair whorling as it curled in opposite directions on combing.

BLOOD GROUPS. Blood groups were identical as established by Dr. Tovey (Regional Transfusion Officer).

TABLE 5

	A	B
Blood Group	B	B
Rh type	Rh ₁ Rh ₂ (CDEc—positive)	Rh ₁ Rh ₂ (CDEc—positive)
M.N.	Type N	Type N

Commentary

The cases are of interest on account of the association of idiocy with gastro-intestinal abnormalities, both apparently genetically determined since they occurred in identical twins. The precise

TABLE 4

	A	B
Motor development :		
4 months' level	4½ successes out of 7 tests	5 successes out of 7 tests
6 months' level	1 success out of 5 tests	2 successes out of 5 tests
9 months' level	2 successes out of 5 tests	3 successes out of 5 tests
Language	No successes beyond 4 months' level	No successes beyond 4 months' level
Adaptive behaviour :		
4 months' level	2 successes out of 3 tests	2 successes out of 3 tests
6 months' level	No successes out of 6 tests	1 success out of 6 tests
9 months' level	No successes	No successes
Social behaviour :		
4 months' level	1 success out of 6 tests No successes beyond 4 months' level	2 successes out of 6 tests No successes beyond 4 months' level

nature of these abnormalities does not appear to coincide with any previously recorded. The nearest approach to cases similar to ours are those described by Joseph Popper (1920), who reported megacolon in twins, one of whom died on the fourth day of its life, the other being diagnosed at the end of four weeks. That there is some degree of megacolon in the present cases is shown by the radiographs, though it is not as extreme as has been found in other cases. On the other hand the patients were resistive when the barium enema was being injected, and probably, but for this, greater dilatation could have been demonstrated. The onset of the condition presumably followed an attack of acute gastro-enteritis at five months, which may indicate some dependence of genetic factors on environmental, i.e. infective. We think that this dependence is much closer than is usually recognized. The cases also have some similarity with that described by Moncrieff and Crichlow (1944) in which dolichocolon was diagnosed by radiograph at ten months and terminated fatally as Hirschsprung's disease at thirty months. The authors remark that their case throws considerable doubt on the usually accepted ideas as to the etiology of Hirschsprung's disease, and especially as to the applicability of the term 'congenital idiopathic dilatation of the colon.' A degree of dolichocolon is to be noted in our cases. Dissimilar features, however, are the occurrence of a partial coeliac syndrome along with considerable gastric dilatation. The coeliac syndrome is indicated by considerable retardation of growth (height 33 in. and 34½ in. respectively in June, 1941, 39 in. and 37½ in. respectively in June, 1947, in contrast to the norms 37 in. at 3¼ years and 51 in. at 9 years), frequent fatty stools, abdominal distension in contrast to the general emaciation of the body, and gluteal wasting. The radiological findings in idiopathic steatorrhea were discussed by Brailsford (1943), who comments on changes in the small intestine and the bones. He reported evidence of irregular tonicity, and a more rapid flow through the intestine so that after two hours barium was seen throughout the whole course of the large intestine. Dilatation of jejunum and large intestine were also noted. Snell and Camp (1934) commented on the smooth contour of the proximal part of the jejunum, which appeared hypertrophied, with clumping of barium in smooth elongated masses. Atrophy and dilatation of the

whole tract were also noteworthy features, though no specific reference was made to gastric dilatation.

Six out of eight cases examined radiologically by Bennett et al. (1932) after the administration of barium enemata showed abnormal dilatation of the colon, from moderate degrees affecting the descending colon only, to an extreme dilatation involving the whole of the large bowel and sufficient to justify the term 'megacolon.' Gastric dilatation, however, does not so far appear to have been described in this connexion.

The occurrence of coeliac disease in identical twins has been noted on four previous occasions during the last fifteen years (Fanconi, 1928; Cervini et al., 1940; Collishaw, 1940; and Holmgren, 1945).

Our sincere thanks are due to Dr. R. M. Norman, Medical Superintendent, Stoke Park Colony, for his kind permission to publish the account of these cases. It is a pleasure to acknowledge his encouragement, helpful criticism, and active interest. Sir Leonard Parsons willingly consented to give us access to his case notes, for which we are deeply indebted. We are grateful to Dr. Tovey of the South-west Regional Transfusion Centre for his expert knowledge and for establishing the identity of the blood groups, and to Mr. A. H. Tingey, M.A., Clinical Pathologist, Stoke Park Colony, for his assistance with the laboratory investigations. We are indebted also to Mr. H. Cox, the radiographer, and to the nursing staff at Stoke Park Colony for their willing co-operation. Mr. T. J. Shields of the British Medical Association Library has perused the previous reports of similar cases, and to him our sincere acknowledgments are also due.

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(For illustrations to this article see Plates V and VI, pp. 257-8.)

(a)—The older twin (A) showing gross abdominal distension, with wasting of the rest of the body. Note the marked gluteal wasting.

(b)—The younger twin (B) showing gross abdominal distension, but with less marked general wasting than in A.

(c)—Radiograph of A showing upward displacement of heart shadow, and of lungs.

(d)—Radiograph of A two hours after barium meal. It shows tremendous dilatation of the stomach.

(e)—Radiograph of A four hours after barium meal, showing 2 loculi in the greatly distended stomach.

(f)—Radiograph of A five and a half hours after barium meal, showing almost empty stomach with considerable distortion in the position of the pyloric outlet and duodenal bulb.

(g)—Radiograph of A after barium enema, showing redundancy and dilatation of the descending colon.

h.—Radiograph of B half an hour after barium meal, showing biloculation of the stomach and gross dilatation. j.—Radiograph of B showing distortion of pyloric outlet and duodenal bulb. k.—Radiograph of B showing marked redundancy and dilatation of colon as far as the splenic flexure.

ACUTE BACTERIAL ENDOCARDITIS BY LAPAGE AND LANGLEY

(1)—Heart, showing hypertrophy of left ventricle, and vegetation on anterior curtain of mitral valve.