"Whistling Face" Deformity in Compound Cranio-facio-corporal Syndrome

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We report a case with an unusual combination of multiple congenital deformities, one of which, the "whistling face" deformity, is extremely rare. The face appears stiff, immobile, and masklike, but no solid mass of muscle is palpable in the buccal region. Burian (1963) reported four cases of a compound cranio-facio-corporal syndrome, but there do not appear to be any other similar reports in the literature.

Case History

The patient, a boy aged 3 years, was brought to the outpatient department by his father on 6 November 1968 because he was unable to open his mouth wide enough to take solid food. The condition had been present since birth and he had been nourished on fluids only. He had been delivered normally at full term, but his mouth was apparently closed by a "membrane." This was removed by a local "medicine man," revealing a narrow mouth opening. The child had puckered lips, which did not relax even during sleep.

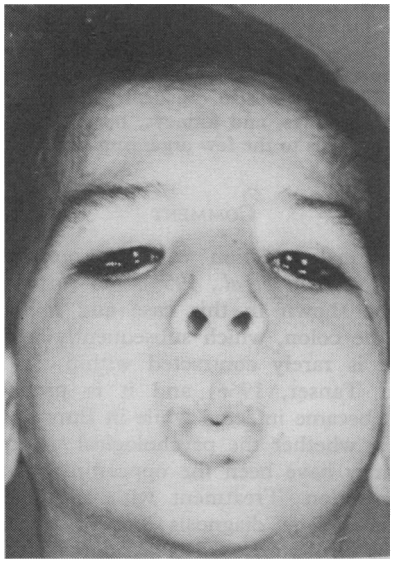

FIG. 1.

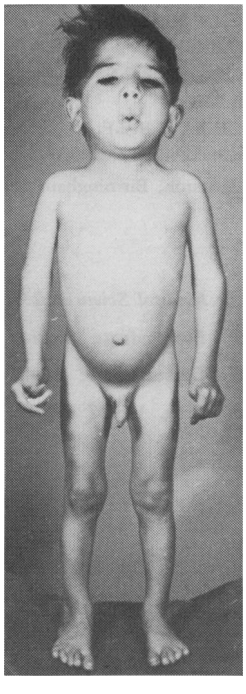

FIG. 2.

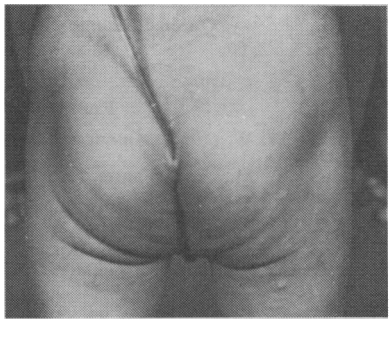

FIG. 3.

Examination showed him to be thin and small for his age. His face looked stiff, immobile, and masklike (Fig. 1). The transverse opening of the mouth was 20 mm. only. There were no scars on the lips, which protruded as in whistling. No muscle mass in the lips or the cheeks could be felt. The palate was high-arched. Speech was unclear and nasal. The teeth were normal for his age. He also had epicanthus and ptosis of both eyelids; alae nasi directed downwards and backwards, with the columella descending backwards to join the upper lip at an obtuse angle; a short, broad neck; ulnar deviation of the third, fourth, and fifth fingers at the metacarpophalangeal joints with contracture of the thumb in adduction in both hands; medial rotation of the third and fourth toes at the metatarsophalangeal joints in the feet; and a postanal dimple (Figs. 2 and 3).

Despite his nasal speech he communicated easily and appeared to be normally intelligent. Electromyographic studies showed minimal activity of the buccinator muscle but normal activity of other facial muscles. Biopsy of the buccinator muscle revealed atrophic muscle fibres.

Operation was undertaken for his puckered mouth. The angles of the mouth were opened by slitting them horizontally after drawing a vertical line from the medial edge of the pupils. The mucosa was advanced outwards and stitched with skin by interrupted chromic catgut sutures. On discharge he was able to open his mouth sufficiently to take solid food.

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Case of Disseminated Histoplasmosis

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Disseminated histoplasmosis rarely causes death, even where the disease is endemic (Addington, 1967). Six cases only of disseminated histoplasmosis have been reported previously in the United Kingdom (Partridge and Tanser, 1966).

Case History

A 63-year-old Caucasian man was first seen as an outpatient in August 1963. He complained of loose and explosive bowel actions for the previous eight months. Before the onset of this disorder he had been fit and well, except for two previous attacks of gastroenteritis, one in 1946, when returning from Burma by troop-ship, and one in 1962, when clinical examination and sigmoidoscopy were negative. A barium enema showed diverticula in the sigmoid colon but no other abnormality. His symptoms then remitted and he remained well until the winter of 1967, when he began to lose weight. At this time he had some personal problems and was drinking heavily and eating little. He was seen again in September 1968 complaining of a 4-st. (25-kg.) weight loss over a period of nine months, six weeks' anorexia and constipation, bilateral ankle swelling, and lassitude of three weeks' duration. On examination he was pale, wasted, and had hepatosplenomegaly and bilateral ankle oedema, more on the right than on the left. There was no lymphadenopathy.

He was admitted to hospital and found to have a hypochromic anaemia. Haemoglobin 10·5 g./100 ml. White cell count and differential normal. E.S.R. 8 mm. per hour. Biochemical studies were not informative. Stool, urine, serum, and a chest *x*-ray picture were normal. Barium meal and follow-through examination

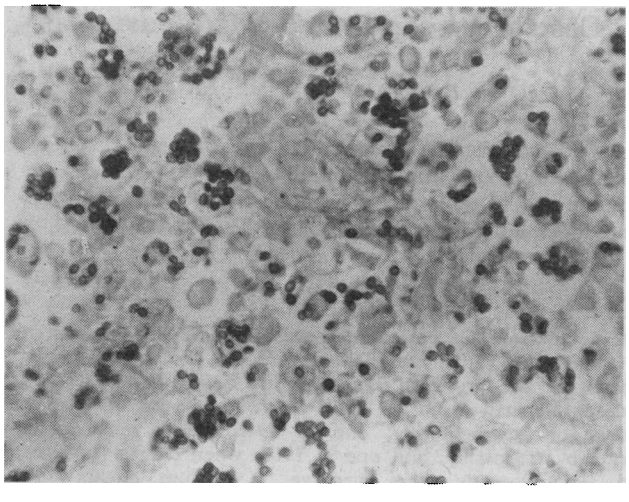
showed the stomach to be displaced by the enlarged liver but no other abnormality. Barium enema findings were essentially the same as those five years previously. An intravenous pyelogram showed an equivocal elongation of the upper calix of the right kidney.

Laparotomy on 17 October showed some clear free fluid in the peritoneal cavity, a large smooth liver with no masses, an enlarged spleen, and a large retroperitoneal mass extending from the upper pole of the right kidney across the great vessels to the left side and down to the splenic flexure of the colon, which was attached to and perforating through into the mass. The tumour was exposed through the lesser sac and found to comprise soft friable white material surrounding an infected cavity communicating with the colon via a 1-cm. perforation at the splenic flexure. Frozen section of the tissue removed from the wall of the abscess showed granulation tissue with numerous histiocytes containing many yeast-like organisms in their cytoplasm. These organisms closely resembled *Histoplasma capsulatum*. This was confirmed on culture. A drainage tube was placed into the cavity and the perforated colon was resected with 5 cm. of normal bowel on either side.

Treatment was begun with amphotericin B 12·5 mg. daily intravenously. (This dose was increased gradually to 50 mg. daily over the next seven days.) Hydrocortisone 25 mg. daily was administered with the amphotericin to reduce side-effects (Tynes *et al.*, 1963). Swabs from the mass showed it to be secondarily infected with *Proteus mirabilis* and *Escherichia coli*, which were sensitive to ampicillin, and he was treated with this drug.

Ten days after operation he collapsed with a massive secondary haemorrhage from the left gastric artery and hilum of the spleen, both sites being involved in an abscess extending from the original mass. The artery was ligated and the spleen removed. At this operation neither the abdominal wound nor the colon anastomosis showed signs of healing and the colon was therefore brought out as a colostomy.

Postoperatively he continued with amphotericin B, hydrocortisone, and ampicillin, together with a high calorie and protein intake intravenously. Alimentary function returned and he was able to take a light but nutritious diet. Despite this his condition continued to deteriorate, and the leg oedema increased to involve the trunk and external genitalia. He developed bilateral blood-stained and infected pleural effusions. He died on 16 November, 30 days after the first laparotomy. During his final illness histoplasma complement fixation and precipitation reactions were negative, but the histoplasmin yeast-phase complement fixation was positive to a dilution of 1/10. Plasma cortisol levels were measured before corticoid therapy and were within normal limits.



High-power view of granulation tissue from colonic perforation containing *Histoplasma capsulatum* within the cytoplasm of the histiocytes and extracellularly. Grocott's silver methenamine technique. (× 450.)

Histology of the resected length of colon showed that the perforation was lined by granulation tissue containing numerous histiocytes laden with *H. capsulatum*, which were also found extracellularly (see Fig.). The spleen was enlarged (455 g.) and contained many small abscesses in which numerous histoplasmas were found, both within the histiocytes and lying free in the central necrotic material. Tissue removed at the second operation consisted of an autonomic ganglion surrounded by granulation tissue laden with histoplasmas.

Necropsy.—At necropsy the surgical wound was poorly healed and there was a large abscess of the right paracolic gutter, which extended across the aorta and inferior vena cava at the level of the adrenal glands. The right gland was part of the wall of the abscess. Histology showed numerous micro-abscesses containing histoplasmas in the left adrenal gland. There was a right subphrenic abscess and an adjacent diaphragmatic pleurisy. A penetrating gastric ulcer 2 cm. diameter was present. Histology of this ulcer showed numerous intracellular and extracellular histoplasmas in the granulation tissue of its walls and base. The liver, 1,850 g., contained a cavernous haemangioma but showed no other focal abnormality. The chest showed no evidence of histoplasma infection other than the right diaphragmatic pleurisy. There was no microscopical evidence of involvement of other systems. Histological examination of the abscess wall showed numerous histoplasmas in the cytoplasm of histiocytes and also extracellularly. There were also organisms in the small vessels of the pancreas, liver, lungs, and kidneys, but there was no sign of reaction in these organs to the few organisms outside the vessels.

COMMENT

Primary intestinal infection with *H. capsulatum*, though rare, occurs (Silverman *et al.*, 1955). No primary pulmonary lesion could be shown in this case and it must therefore have been in the colon, which subsequently perforated.

This disease is rarely contracted within the British Isles (Partridge and Tanser, 1966) and it is probable that the present patient became infected while in Burma. It is interesting to speculate whether the psychological upset at the onset of his illness may have been the opportunity for reactivating the original infection. Treatment with amphotericin B was begun immediately the diagnosis had been made. Though this therapy has been successful (Partridge and Tanser, 1966), it proved ineffectual in this case, and at necropsy many histoplasma organisms were found in tissues throughout the body.

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