Preliminary Communications

Maple Syrup Urine Disease

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Maple syrup urine disease is a biochemical rarity (Menkes, Hurst, and Craig, 1954; Dancis, Levitz, and Westall, 1960). In the following case some biochemical findings are different from those previously reported.

CASE REPORT

A full-term female child was well for two weeks after birth but then became very drowsy. At 1 month her mother noticed a peculiar odour. At 6 weeks the health visitor thought the "phenistix" test was positive. The parents are not consanguineous and have two healthy older daughters. A female child born in 1960, who died of

Serum amino-acid chromatogram. Note the intense leucine/isoleucine and valine spots.

bronchopneumonia at 3 weeks, was drowsy from birth and fed poorly.

Examination showed our patient to be unresponsive, with increased tone and occasional opisthotonic spasms. A strong odour of maple syrup was present.

Investigations.—Urinary chromatograms showed marked increase in valine (from which the odour was detected), leucine, and isoleucine, with moderate increases in other amino-acids. The α -keto acids of the branched amino-acids were also present in large amounts. Indolyl-lactic acid was present in excess but not indolyl-acetic acid. Asparagine and ethanolamine were also detected. The serum and cerebrospinal fluid showed greatly increased amounts of valine, leucine, and isoleucine; other amino-acids were normal or low in quantity (see Fig.).

COMMENT.—While clinically this case is one of maple syrup urine disease the biochemical findings suggest the presence of a renal tubular defect and hepato-cellular damage. Previously reported cases have shown an increase in indolyl-acetic acid; this was not present in the above case. Detailed biochemical findings will be published later, with the results of a dietary trial which is proceeding.

We are grateful to Mr. J. T. Ireland (Alder Hey Children's Hospital, Liverpool) for technical help; to Mr. Milner, of Scientific Hospital Supplies, for help with dietary regime; and to Mrs. M. F. Down for clerical assistance.

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Medical Memoranda

Ruptured Abdominal Aortic Aneurysm in Marfan's Syndrome

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Marfan's syndrome is an ill-defined defect of connective tissue, usually hereditary, manifest by multiple abnormalities involving in particular the skeleton, the eyes, and the cardiovascular system.

When the aorta is involved, aneurysmal dilatation occurs usually in the ascending part with severe associated aortic insufficiency (McKusick, 1960; Muller, Dammann, and Warren, 1960). Aortic lesions that are more distal also occur (Austin and Schaefer, 1957; Van Buchem, 1958; Roark, 1959; McKusick, 1960). Aneurysm of the abdominal aorta in

Marfan's disease has been described in at least nine cases (Davis, Benson, and Miller, 1957; Van Buchem, 1958; Hardin, 1959; Sinclair, Kitchin, and Turner, 1960; McKusick, 1960; Killip and Holmquist, 1961). Rupture of the aneurysm occurred in three of these cases, two of which were treated surgically. None of these three cases survived.

CASE REPORT

A 24-year-old white male student developed severe abdominal pain on 6 July 1961 after swimming while on holiday in Northern Ireland. He was transferred to the Royal Victoria Hospital, Belfast, next day. On admission he was in a state of shock, with very severe pain in the back, the lower abdomen, and the right hip and scrotum. There was a large pulsatile swelling in the central and upper abdomen. The femoral pulses were both present. It was noted that he was unusually tall and thin with a long, narrow, aquiline face, blue sclera, and bilateral dislocated lenses. The palate

was markedly arched and the voice high-pitched. The fingers and toes, especially the great toes, were unusually long and slender. In 1957 a diagnosis of ankylosing spondylitis had been made, for which he had received a course of deep x-ray therapy.

A diagnosis of ruptured abdominal aortic aneurysm was made. A blood transfusion was started and operation was carried out under hypothermia (30° C.) through a long left paramedian incision extended into the left side of the chest. A large mottled blue pulsatile swelling was found extending over most of the posterior abdominal wall concealing the aorta and overlapping the common iliac vessels. The lower thoracic aorta was now occluded with a tape, the common iliacs controlled with bulldog clamps, and the aneurysmal sac opened. After removing the clot it was possible to apply an arterial clamp just above the upper limit of the aneurysm and immediately below the renal vessels. The aneurysm was excised except for a strip of the posterior wall beside the vena cava. It was noted that the posterior wall of the aorta below the renal vessels was very thin and the left iliac vessels were twice the size of those on the right side. A Y-shaped, woven, crimped "teflon" graft was sutured above to the aorta and below to the common iliac arteries.

After the operation the patient's progress was uneventful apart from some abdominal distension during the first 24 hours.

Sections from the excised portion of aorta and common iliac vessels showed typical changes of advanced cystic medionecrosis.

Post-operative History.—The patient returned to England at the end of July 1961 and was able to resume work. In May 1962 his blood pressure was 145/90 and all peripheral pulses were present. A radiograph of his chest showed no abnormality. His measurements were: height, 6 ft. 2½ in. (1.9 m.); "lower segment" (upper border of the pubis to the floor), 3 ft. 2 in. (1 m.); "upper segment," 36½ in. (0.9 m.); span, 6 ft. 2½ in. (1.86 m.); metacarpal index (calculated from a radiograph of the right hand), 8.9. (Normal range: 5.4–7.9 (Sinclair *et al.*, 1960).)

Aorta, showing extensive mucoid degeneration of media with loss of elastic tissue and muscle. (H. and E. ×300.)

In June 1962, after an attack of biliary colic, he was found to have gall stones. A cholecystectomy was carried out by Mr. R. O. Lee at Northampton General Hospital. At this operation the three anastomoses of the graft were patent and all lower limb pulses were palpable.

In December 1962 he developed claudication in the left leg. Mr. H. H. G. Eastcott found that the left limb of the graft had throm-

bosed. He excised the lower part of the graft and replaced it with a knitted "dacron" prosthesis. The patient made a good recovery from this operation.

Family History.—The family history has been traced back both to maternal and paternal grandparents. There is no definite evidence of any of them having any of the stigmata of Marfan's syndrome.

COMMENT

Sinclair *et al.* (1960) stress the frequency with which patients suffering from Marfan's syndrome have musculo-skeletal complaints. Seven of their 40 patients had low back pain, and in three of these patients a diagnosis of ankylosing spondylitis had been made erroneously. A similar misdiagnosis was made in our patient, as radiographs of the spine and pelvis in 1961 showed no abnormality. Back pain in Marfan's syndrome is usually due to skeletal abnormalities, the commonest being kyphoscoliosis. A rarer and more serious cause is an expanding aortic aneurysm (Hancock and Irwin, 1962).

The hazards of operative treatment for aneurysm of the aorta in Marfan's syndrome are considerable. The wall of the aorta may be like "layered wet blotting-paper" (Hurley, 1959). Cooley (1960) states that sutures tear out as they are inserted and disruption may occur several hours or days later. However, Davis, Benson, and Miller (1957) and Dubost (quoted by McKusick, 1960) have successfully replaced abdominal aortic aneurysms in Marfan's syndrome by grafts. Baker, Wilson, and Woods (1958) successfully replaced an aneurysm of the descending aorta, and Muller *et al.* (1960) operated with success on three cases of aneurysm of the ascending aorta in Marfan's syndrome. In our case success attended resection for a ruptured abdominal aortic aneurysm, but the posterior aortic wall at the proximal level of resection just beyond the renal vessels was grossly involved. This patient has since survived excision and replacement of the lower half of the graft, but his long-term prognosis remains poor.

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