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.

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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