CASE REPORT

TWO CASES OF GLYCOGEN DISEASE

BY

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In 1929, von Gierke described two cases of a previously undefined condition which he called 'Hepatonephromegalia glycogenica.' In a paper in 1931 he used the name 'Thesaurismosis glycogenica' and this term has since been used by Spanish authors (Jiménez Diaz et al., 1940). It is now usually called 'Glycogen disease,' 'Glycogen storage disease,' or 'von Gierke's disease.' Cases probably of this condition had been described previously by Wagner and Parnas (1921) and Snapper and van Creveld (1928). Full reviews of the subject have been published by Ellis and Payne (1936), van Creveld (1939), and Atkinson (1939).

Two cases are here described. An earlier report on the first was made by Warner (1933).

Case 1. Female, aged eight years.

HISTORY. The child was first seen at the age of two years with jaundice and an enlarged abdomen. For ten days she had had a cough and for seven days mild jaundice. The stools were white and the urine dark orange coloured. The liver extended $\frac{1}{2}$ inch below the umbilicus. There were two dilated veins on the abdominal wall.

The jaundice passed off gradually and the liver was found to extend to 1 inch above the umbilicus. Since then the liver has gradually become larger and she has had repeated attacks of bronchitis.

At the age of six-and-a-half years, her height was 39 $\frac{1}{2}$ inches; weight 38 lb. 8 oz.; mentality normal.

The liver was uniformly enlarged 2 inches below the umbilicus at the outer border of the rectus abdominis; it was firm but not hard. The spleen was not palpable. Free fluid was never found in the abdomen.

FAMILY HISTORY. One sibling died and is said to have had a large liver; one other is alive and well.

PATHOLOGICAL INVESTIGATIONS. Age 3 years—

Blood W. R. negative.

Blood W. R. of both parents negative.

Van den Bergh reaction—direct—negative.
indirect—less than 0.5 units.

Faeces—wet film normal.

Age 5 years: Blood count: Red blood cells 4,920,000 per c.mm.; Hb. 80 per cent.; Colour index 0.8; White blood cells 6,800 per c.mm.; polymorphs. 52 per cent.; lymphocytes 46 per cent.; large mononuclears 2 per cent.

Blood sugar curves $\left\{ \begin{array}{l} 0. \text{ 86 mgm. per cent.} \\ \text{Hours } \left\{ \begin{array}{l} 1. \text{ 160 mgm. per cent.} \\ 2. \text{ 152 mgm. per cent.} \\ 4. \text{ 112 mgm. per cent.} \end{array} \right. \end{array} \right.$

Urine—sugar absent and acetone present.

Blood sugar after injecting minims 4 of liq. adrenalin.:

1. 48 mgm. per cent.
2. 50 mgm. per cent.
3. 44 mgm. per cent.
4. 44 mgm. per cent.

Control

1. 69 mgm. per cent.
2. 100 mgm. per cent.
3. 94 mgm. per cent.
4. 89 mgm. per cent.

Urine: sugar and acetone absent.

Age 7 years: Stools contained 14 per cent. starch of dried weight.

Age 8 years: Blood count: Red blood cells 3,060,000 per c.mm.; Hb. 38 per cent.; White blood cells 9,200 per c.mm.

In six weeks the haemoglobin rose to 74 per cent.

Case 2. Male, aged seven years.

HISTORY. Since the age of four years he has had a large abdomen with abdominal pain on hurrying. He eats well, does not vomit and has his bowels open regularly.

PAST HISTORY. Diphtheria, measles and bronchitis. He is said to catch cold easily. He had never been jaundiced.

FAMILY HISTORY. He is an only child.

ON EXAMINATION. Age six years. Weight 41 lb. The liver was $3\frac{3}{4}$ inches below the costal margin at the outer border of the right rectus abdominis; the spleen was not palpable.

Age seven years. Weight 44 lb. The lower border of the liver was at the level of the umbilicus.

PATHOLOGICAL INVESTIGATIONS. Blood count: Red blood cells 4,500,000 per c.mm.; Hb. 86 per cent.; Colour index 0.95; white blood cells 4,000 per c.mm.; polymorphs 49 per cent.; lymphocytes 46 per cent.; eosinophils 3 per cent.; large mononuclears 2 per cent.

Blood W.R. negative.

Van den Bergh reaction: direct and indirect negative.

Blood sugar curve:

0. 120 mgm. per cent. urine—sugar negative, acetone negative.
1. 134 mgm. per cent.
2. 153 mgm. per cent. urine—sugar negative, acetone positive.
3. 157 mgm. per cent.
4. 166 mgm. per cent. urine—sugar negative, acetone positive.

Blood sugar curve after injection of adrenaline 117/111/122/117 mgm. per cent.

Sugar curve after giving test dose of laevulose: 76/92/73/73/73 mgm. per cent.

Galactose tolerance test: 20 gm. of galactose were given by mouth and specimens of urine were collected hourly for five hours. During this period no galactose was excreted in the urine. Normally at least 2 gm. of the sugar should have been excreted.

The dried stool contained 0.7 per cent. of carbohydrate.

Discussion

The two cases described are typical examples of hepatomegalic glycogen disease. They illustrate three points that have received comparatively little attention:

1. The presence of starch in the faeces.
2. The occurrence of the disease in siblings.
3. A past history of jaundice.

STARCH IN THE FAECES. Naish and Gumpert (1935, 1936) described a typical case of glycogen disease in whose stools an enormous quantity of starch was visible microscopically. On analysis the fat content was found to be normal but the dried stool contained 34 per cent. of starch (estimated as glucose). The faeces were three times as potent as those of a normal child in digesting a 1 per cent. starch solution and the diastatic activity remained unchanged after removing all the organisms. The faeces were strongly acid, and after neutralization, fermentation of the starch took place *in vitro*. These observations appear to have attracted little attention and references to faecal starch are rare in descriptions of cases of this disease. Wagner and Parnas (1921) mention fermentation of the stool in their patient. Van Creveld (1939) found no starch in the faeces of one of his patients. Case 1 had 14 per cent. of starch in the dried stool whereas case 2 had only 0.7 per cent.

FAMILY HISTORY. Although there is no family history of the disease in the majority of cases it is becoming realized that it may occur in siblings.

Such cases have been described by Ellis and Payne (1936) and Bauzá (1935, quoted by Atkinson, 1939) and the cardiomegalic type by Sprague, Bland and White (1931). Probable examples have been reported by Unshelm (1932), Beltingham-Smith and O'Flynn (1933), Lindsay, Ross and Wigglesworth (1935), Krakower (1936) and Antopol et al. (1940); case 1 would come under this heading. Exchaquet (1931) described an interesting family, consisting of a girl aged twelve and twins, a boy and a girl, aged three. All were underdeveloped and had had large livers and ketonuria since birth. The fasting blood sugars were normal but the sugar tolerance curves were abnormal. Sugar tests of liver function were normal but urobilin and urobilinogen were present in the urine and the blood uric acid was raised. The diagnosis of these children's illness remains unproved.

PAST HISTORY OF JAUNDICE. A previous history of jaundice has usually been regarded as an independent illness unconnected with glycogen disease but a number of cases similar to case 1 have been reported (Anderson, 1935; Ellis and Payne, 1936; Sundal, 1936; Gardiner and Simpson, 1938; Mason and Anderson, 1941). It seems likely that the jaundice is an integral part of the illness.

Summary

Two typical cases of the hepatic type of glycogen disease are described. In one of them a sibling died with an enlarged liver, there was a past history of jaundice and large quantities of starch were present in the stool.

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