Lupus erythematosus cells obtained from the patient.

T. LEHOCZKY *ET AL.*: SKELETAL MUSCLE GLYCOGENOSIS IN IDENTICAL TWINS

FIG. 1

FIG. 2

FIG. 3

FIG. 1.—Haematoxylin and eosin stain ; cross-section, low-power view.

FIG. 2.—Same as Fig. 1, high-power view.

FIG. 3.—Hale's method and P.A.S. stain (combined with hyaluronidase) ; cross-section, high-power view. Arrows indicate the mucopolysaccharide.

overlying scalp laceration may be small and neurological symptoms at first not in evidence. The more serious underlying damage may thus be overlooked. It leads one to emphasize again that all scalp lacerations must be explored thoroughly. Where a history of a blow on the head with a stiletto heel is obtained particular care must be exercised.

I am indebted to Mr. A. Sutcliffe Kerr and Mr. C. B. Sedzimir for permission to publish details of their cases, and to the latter for much helpful criticism and advice.

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Skeletal Muscle Glycogenosis in Identical Twins

[WITH SPECIAL PLATE]

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Glycogen-storage diseases can be divided into at least six types (Stetten and Stetten, 1960), in two of which muscle disorders are a striking feature.

This report concerns two 29-year-old male twins presenting with a somewhat atypical form of muscular glycogenosis. Complete biochemical, histological, and histochemical investigations were possible in one twin only, but the history, clinical features, and effort tests for ischaemia were similar in both.

Each patient presented with a history extending over one or two years of gradual and then severe muscle fatigue, limb pains, with cramp of the muscles after any rhythmical exertion. Fatigue was determined by various muscle exercises and included the ischaemic exercises already described (McArdle, 1951; Schmid and Mahler, 1959; Pearson, Rimer, and Mommaerts, 1961). These latter experiments demonstrated fatigue and exhaustion much sooner in the presence of ischaemia than in its absence. Neurological examination did not reveal any other abnormalities.

One twin was admitted to hospital on three occasions within seven months, during which time the following detailed investigations were performed.

Biochemical Studies.—The resting serum pyruvic acid was raised to 3.1 mg./100 ml.; in the second minute of the ischaemia-effort experiment and while in complete exhaustion it rose to 5 mg./100 ml., which is twice the normal value. The resting serum lactic acid was 20 mg./100 ml. (upper limit of normal), and during ischaemia it was 24 mg./100 ml. Normal controls tested in a similar manner gave lactic acid figures which showed a fourfold to fivefold increase over the resting level. These findings indicated that anaerobic muscular glycolysis was partially inhibited.

Histological Studies.—Biopsy specimens were obtained from the quadriceps femoris muscle of each leg, and both muscles showed identical lesions. Sections stained with haematoxylin and eosin, haematoxylin and van Gieson, and by Mallory's method all showed the presence of numerous vacuoles of varying size in the muscle (Special Plate, Figs. 1 and 2), seen both in transverse and in longitudinal section. There was also a moderate proliferation of sarcolemmal nuclei.

Staining with Sudan III and oil red O did not stain the vacuoles, but Best's carmine stain disclosed an increased glycogen content of the muscle; staining with periodic acid Schiff (P.A.S.) after Lison's fixation as well as with Hale and P.A.S. stain combined with hyaluronidase demonstrated the

presence of mucopolysaccharide (Special Plate, Fig. 3). Normal amounts of phosphorylase were found using the method of Takeuchi.

DISCUSSION

These cases seem to differ from the classical type of glycogen disease of striate muscle in showing a slight rise in blood lactate on exercise as well as demonstrating a normal muscle phosphorylase content. Thus these patients differ from the cases in which serum lactic acid and pyruvate are not increased (Pearson *et al.*, 1961; Schmid and Hammaker, 1961; di Sant'Agnese, Andersen, and Metcalf, 1962), although they more closely resemble other recorded cases showing a slight increase (Schmid and Mahler, 1959; Mellick, Mahler, and Hughes, 1962). Our own experience suggests that a slight rise in lactic acid in the serum indicates a glycogen myopathy.

Vacuoles in the muscle fibres have been recorded previously in similar cases of myopathy (Wolff, 1936; Schnabel, 1958) and in most cases glycogen has been demonstrated as it was in our patient (Zellweger, Dark, and Abu Haidar, 1955; Schmid and Mahler, 1959; Pearson *et al.*, 1961; Thomson, MacLaurin, and Prineas, 1963). In some similar myopathies a normal or a diminished glycogen content has been found in the vacuoles (Mellick *et al.*, 1962; Engel, 1962). Acid mucopolysaccharide in the vacuoles in the muscle fibres has been recorded only once previously (Zellweger *et al.*, 1955).

In McArdle's disease phosphorylase is absent (McArdle, 1951; Pearson *et al.*, 1961; Mellick *et al.*, 1962; Engel, 1962; Thomson *et al.*, 1963). However, in the second case reported by Thomson *et al.* (1963) muscle phosphorylase was normal.

It would appear that the case recorded here is similar to the child of 4 years recorded by Thomson *et al.* (1963) and would be placed in type 5 of the classification of Stetten and Stetten (1960), where the enzyme deficiency is unknown.

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REFERENCES

- di Sant'Agnese, P. A., Andersen, D. H., and Metcalf, K. M. (1962). *J. Pediatr.*, **61**, 438.
Engel, W. K. (1962). *Neurology (Minneapolis)*, **12**, 778.
McArdle, B. (1951). *Clin. Sci.*, **10**, 13.
Mellick, R. S., Mahler, R. F., and Hughes, B. P. (1962). *Lancet*, **1**, 1045.
Pearson, C. M., Rimer, D. G., and Mommaerts, W. F. H. M. (1961). *Amer. J. Med.*, **30**, 502.
Schmid, R., and Hammaker, L. (1961). *New Engl. J. Med.*, **264**, 223.
— and Mahler, R. (1959). *J. clin. Invest.*, **38**, 2044.
Schnabel, R. (1958). *Virchows Arch. path. Anat.*, **331**, 287.
Stetten, D., jun., and Stetten, M. R. (1960). *Physiol. Rev.*, **40**, 505.
Thomson, W. H. S., MacLaurin, J. C., and Prineas, J. W. (1963). *J. Neurol. Neurosurg. Psychiat.*, **26**, 60.
Wolff, K. (1936). *Beitr. path. Anat.*, **97**, 289.
Zellweger, H., Dark, A., and Abu Haidar, G. A. (1955). *Pediatrics*, **15**, 715.