RIBOSURIA IN MUSCULAR DYSTROPHY

BY

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Ribosuria as a characteristic feature of muscular dystrophy was reported by Minot, Frank, and Dziewiatkowski (1949). The ribosuria was detected by a positive reduction with Benedict's qualitative reagent on prolonged heating and the sugar identified by the preparation of its osazone. A test for muscular dystrophy based on the microscopical appearance of this osazone was devised by Orr and Minot (1952). It was claimed that this reaction for ribose was positive in all cases of progressive muscular dystrophy, dystrophia myotonica, myotonia congenita and amyotonia congenita, and in some cases of poliomyelitis in the febrile stage, but was negative in a variety of other conditions such as myasthenia gravis and progressive muscular atrophy.

We therefore examined the urine of patients suffering from a variety of myopathic disorders for ribose by a sensitive and specific paper chromatographic technique.

Methods

Urine Specimens.—Twenty-four hour collections were made from ward patients and early morning specimens obtained from out-patients; no preservative was employed. Each urine was tested by heating 8 drops for 45 minutes in a boiling water-bath with 5 ml. of Benedict's qualitative reagent and by the paper chromatographic technique described below. These tests were performed on each specimen when it was received in the laboratory and after it had been allowed to stand for 24 hours at room temperature. The two tests were repeated after the addition of 20 ml. concentrated HCl and a further 24 hours' standing.

Paper Chromatographic Method for Ribose.—A descending method using one dimension strips of Whatman No. 1 filter paper and a n-butanol-acetic acid-water mixture was employed (Horrocks and Manning, 1949). Each chromatogram was run for 24 hours and the spraying reagent was aniline hydrogen phthalate (0.9 ml. of redistilled aniline dissolved in 100 ml. of 0.1M-phthalic acid). Glucose and ribose were used as marker substances, the Rf values being 0.19 and 0.33 respectively (uncorrected for temperature). The spraying reagent gave a yellow-brown spot with glucose and a red-brown spot

with ribose. The spots exhibited intense greenish-blue fluorescence under ultra-violet light (Consden and Stanier, 1952). A "hanovia chromalite" lamp was used as the light source. The intensity of this fluorescence was such that it was easily possible to detect 5 µg. of the sugars on the chromatograms. Quantities of urine up to 100 µl. were used in each run so that the sensitivity of the method was at least 5 µg. in 100 µl. of urine. Therefore if no ribose was detected on the chromatogram the urine specimen must have contained less than 5 mg. ribose per 100 ml. Assuming an average daily urine volume of 1,500 ml. less than 75 mg. in 24 hours of ribose were excreted.

CLINICAL MATERIAL

Condition	Type	No. of Cases
Progressive muscular dystrophy	(a) Pseudohypertrophic muscular dystrophy	1
	(b) Facioscapulohumeral muscular dystrophy	1
	(c) "Late forms"	4
Dystrophia myotonica		2
Ocular myopathy	Kiloh and Nevin (1951)	1
Poliomyelitis	Febrile stage	2

Results and Discussion

In only two urines out of 11 examined was any reduction obtained on testing with Benedict's qualitative reagent. In no case was ribose detected on a paper chromatogram and therefore less than 5 mg. per 100 ml. of ribose was present in the urine samples. The two specimens which reduced Benedict's qualitative reagent and were from patients with muscular dystrophy and poliomyelitis respectively, when chromatographed on paper showed the presence of glucose only. The failure to demonstrate ribose, even after the urine was allowed to stand, is important because Minot and others (1949) considered that in cases of muscular dystrophy the ribose was excreted as a phosphate complex which became converted to free ribose on standing.

The results show that the excretion of significant amounts, i.e., more than 5 mg. per 100 ml., of ribose

in the urine did not occur in the patients we have studied. We do not consider therefore that ribosuria is a characteristic feature of muscular dystrophy and allied diseases, and a test based on its alleged association with these conditions is of no value in differential diagnosis.

Summary

The urine of patients suffering from muscular dystrophy and certain allied conditions has been examined for ribose by a sensitive and specific paper chromatographic technique. Ribose was not detected in the urine specimens and it is concluded

that ribosuria is not a characteristic feature of this group of diseases.

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