THYROTOXICOSIS IN TWO ADULT MONGOLS

By

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ABSTRACT

Two cases of thyrotoxicosis in adult mongols are reported. This combination appears to contradict the theory that mongolism is due to pituitary hypofunction resulting in hypothyroidism. The combination is most likely coincidental.

Gilchrist (1946) described what was probably the first case of thyrotoxicosis in a mongol. It seems that only five cases with this combination have been reported, two of which were in adults (Gilchrist 1946; McGirr & Murray 1956; Esen & Mautner 1957; H. Andersen, pers. comm.; Nickey 1960).

MATERIAL AND METHODS

The present report includes two cases of thyrotoxicosis in adult mongoloid women.

In addition to a complete physical examination of the patients and a full record of the usual routine laboratory examinations basal metabolic rate, the excretion of $^{131}$I in the urine 6–24 hours after administration and the cholesterol in the serum were investigated.

Stoa (1953) found 13–35 % of the dose given as normal limits for excreted $^{131}$I in the urine.

Cholesterol in serum was investigated by the method of Klungsøyr et al. (1958). The results by this method are approximately 14 % higher than those obtained by the method of Schoenheimer-Sperry (1934).

Case 1. Female aged 41 years. There are no other known cases of thyroid disease or mongolism in the family. The mother was 40 years of age when the patient was born. The patient has two elder siblings who are well. She went to a special school and can write intelligible letters and hold a simple conversation.

Six years before admission she weighed 75 kg, but during the last 6 months in particular she had lost weight though her appetite remained unchanged. On admission on 28.7.59 her weight was 53 kg, height 151 cm. During the months immediately
before admission she developed a goitre and protuberant eyes; in addition she was very inclined to sweat. Her basal metabolic rate when ambulant was 135%. She was of mongoloid appearance. Both little fingers were abducted and short with an ulnar convexity. The thumbs were short, and the ears small. There was a large gap between the first and second toes, particularly on the right foot.

The thyroid gland, particularly the left lobe, showed considerable diffuse enlargement. In addition there was exophthalmos, a positive Graefe's sign and hot, smooth, clammy skin. She appeared to be restless and nervous. Blood pressure 150/60. Pulse 120 regular. ECG: sinus tachycardia. A grade III systolic murmur was heard over the heart. This was loudest at the apex. Basal metabolic rate: 129%. 131I excretion in the urine 6–24 hours after administration: 8.7% of the dose given. Cholesterol: 228 mg/100 ml, 219 mg/100 ml.

X-ray investigation showed slight deviation of the trachea to the right and some calcification in the thyroid gland.

The patient was treated with 1-methyl-2-mercapto-imidazole. After 1½ years the dose was reduced to 5 mg every other day.

Case 2. Female aged 21 years. There are no other cases of mongolism in the family. The mother had two abortions before the birth of the patient. She was 31 years of age when the girl was born. The father has a non-toxic goitre, and goitre is common in the district in which the patient was brought up. The patient is an only child. She did not go to school, but helped a little in the house.

The patient was previously overweight, but during the last 1½ years before admission on 27.6.58 she lost 9 kg even though her appetite increased. At the same time exophthalmos developed. She was troubled by sweating, nervousness and slight dyspnoea on exertion.

The patient had a typical mongoloid appearance. She was not able to make herself understood, and she spoke indistinctly with a stammer. The skin was hot and clammy; there was tremor of the hands and pronounced exophthalmos. The thyroid gland showed moderate diffuse enlargement and there was a grade II systolic murmur over the apex of the heart. Pulse 96. Blood pressure 125/70. ECG: sinus tachycardia. On X-ray the heart and the trachea were normal. Basal metabolic rate: 136%, 127%. 131I excreted in the urine 6–24 hours after administration: 8.7%. Cholesterol: 102 mg/100 ml.

The patient was treated with 4 mc 131I. On follow-up three months later, the thyrotoxic symptoms had to some extent regressed. The weight had increased from 50.7 to 51.8 kg. Basal metabolic rate: 124%, 117%. 131I excretion in the urine 6–24 hours after administration: 16.5% of the dose given. Cholesterol: 103 mg/100 ml. The patient was considered to be slightly thyrotoxic, and was treated with 1-methyl-2-mercapto-imidazole.

**DISCUSSION**

Mongolism is said to occur as the result of an injury to the embryo in the 6–14th week of intrauterine life (Benda 1949 b; Ingals 1947). The deleterious factor is either of a genetic nature or is present in the embryo's environment (Benda 1949 b). Benda (1949 a) considers that the function of the thyroid gland is reduced in mongolism due to lack of thyrotrophic hormone. The cases of definite thyrotoxicosis in mongols that are on record do not support this theory.
On investigating the uptake of radioactive iodine in mongols Friedman (1955) found that there was no significant difference between the uptake in mongols and that in a control group of euthyroid, mentally subnormal subjects. He concluded that it is not thyroid disease that leads to the development of a mongoloid child.

In the last few years, the genetic nature of mongolism has been demonstrated by investigation of the chromosomes in mongols (Lejeune et al. 1959; Jacobs et al. 1959).

The occurrence of hyperthyroidism or hypothyroidism in a mongol is probably coincidental.

REFERENCES

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