Skull Changes in Eighteen Cases of Dystrophia Myotonica

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SKULL CHANGES IN EIGHTEEN CASES OF
DYSTROPHIA MYOTONICA

by

Giovanni Di Chiro and John Egerton Caughey

Dystrophia myotonica (myotonica atrophica) is a heredofamilial disorder which is inherited as an autosomal dominant. When the disease first appears in a family, it presents with cataract. This may be the only detectable evidence of the disease in one or two or maybe three generations. Usually, however, in the second or so-called dystrophic generation, the cataract develops at an earlier age and other features of the fully developed disorder appear, such as myotonia, muscle wasting, gonadal atrophy, frontal baldness, and cardiac abnormalities of conduction and rhythm.

Few authors have referred to the radiographic changes in the skull in these patients. Scharnke and Full (1920) reported in one case large frontal sinuses with a normal pituitary fossa. Rouques (1931) described the skull roentgenographic findings in four patients with dystrophia myotonica but did not emphasize this as an aspect of the disorder. In his cases there was thickening of the calvaria in three, in two the sella turcica was small, in two the frontal sinuses were large, and in two there was a bridged sella. Fagin (1946) reported hyperostosis frontalis in one male patient. One of us (Caughey & Brown 1950) in an endocrine study of nine cases of dystrophia myotonica reported on the radiographic skull changes and suggested that these should come to be

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Table

<table>
<thead>
<tr>
<th>Patient</th>
<th>Age and Sex</th>
<th>Approximate duration of illness in years</th>
<th>Gonadal changes if present</th>
<th>General shape</th>
<th>Vault</th>
<th>Paranasal sinuses</th>
</tr>
</thead>
<tbody>
<tr>
<td>D.J.</td>
<td>35 ♀</td>
<td>9</td>
<td>Tubular atrophy</td>
<td>0</td>
<td>Thickened calvaria, hyperostosis frontalis interna</td>
<td>Large sphenoidal</td>
</tr>
<tr>
<td>C.H.</td>
<td>35 ♂</td>
<td>13</td>
<td></td>
<td>0</td>
<td>Thickened calvaria in frontal region</td>
<td>Frontal moderately large</td>
</tr>
<tr>
<td>D.H.</td>
<td>44 ♂</td>
<td>5</td>
<td></td>
<td>0</td>
<td>Thickened calvaria in frontal region</td>
<td>Frontal moderately large</td>
</tr>
<tr>
<td>C.F.</td>
<td>18 ♂</td>
<td>10</td>
<td></td>
<td>0</td>
<td>Thickened calvaria</td>
<td>Large sphenoidal</td>
</tr>
<tr>
<td>E.W.</td>
<td>36 ♂</td>
<td>16</td>
<td>Small testicles, tubular atrophy</td>
<td>Acromegaloïd</td>
<td>Thickened calvaria</td>
<td>Frontal very large</td>
</tr>
<tr>
<td>F.K.</td>
<td>37 ♂</td>
<td>25</td>
<td>FSH &gt; 172</td>
<td>0</td>
<td>Thickened calvaria</td>
<td>Frontal moderately large</td>
</tr>
<tr>
<td>P.K.</td>
<td>11 ♂</td>
<td>1</td>
<td>Hysterectomy and oophorectomy at 38</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>T.L.</td>
<td>29 ♂</td>
<td>1</td>
<td>Acromegaloïd</td>
<td>0</td>
<td>Thickened calvaria</td>
<td>Large frontal</td>
</tr>
<tr>
<td>J.W.</td>
<td>44 ♀</td>
<td>30</td>
<td>Hystereotomy and oophorectomy at 38</td>
<td>0</td>
<td>Thickened calvaria</td>
<td>Frontal absent</td>
</tr>
<tr>
<td>V.W.</td>
<td>47 ♀</td>
<td>20</td>
<td>Hyperostosis frontalis interna</td>
<td>0</td>
<td>Hyperostosis frontalis interna</td>
<td>0</td>
</tr>
<tr>
<td>G.W.</td>
<td>41 ♂</td>
<td>17</td>
<td>Small testicles (No biopsy)</td>
<td>Acromegaloïd</td>
<td>Thickened calvaria</td>
<td>Large frontal</td>
</tr>
<tr>
<td>M.T.</td>
<td>33 ♀</td>
<td>15</td>
<td></td>
<td>0</td>
<td>Thickened calvaria</td>
<td>0</td>
</tr>
<tr>
<td>R.V.</td>
<td>22 ♂</td>
<td>4</td>
<td>Large frontal</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>L.W.</td>
<td>39 ♂</td>
<td>3</td>
<td>Small testicles, tubular atrophy</td>
<td>0</td>
<td>Thickened calvaria</td>
<td>0</td>
</tr>
<tr>
<td>F.W.</td>
<td>49 ♀</td>
<td>4</td>
<td>Tubular atrophy</td>
<td>0</td>
<td>Thickened calvaria, hyperostosis frontalis interna</td>
<td>Small frontal</td>
</tr>
<tr>
<td>I.B.</td>
<td>35 ♀</td>
<td>15</td>
<td>FSH &gt; 96</td>
<td>0</td>
<td>Thickened calvaria</td>
<td>0</td>
</tr>
<tr>
<td>J.W.</td>
<td>33 ♀</td>
<td>15</td>
<td>Hyperostosis frontalis interna</td>
<td>0</td>
<td>Hyperostosis frontalis interna</td>
<td>Small frontal</td>
</tr>
<tr>
<td>J.U.</td>
<td>48 ♂</td>
<td>2</td>
<td>Tubular atrophy</td>
<td>0</td>
<td>Hyperostosis frontalis interna</td>
<td>Large frontal</td>
</tr>
</tbody>
</table>

"0" indicates within normal limits.

accepted as one of the variable features of the disorder. In subsequent papers Caughey (1952) added further case reports. Among twenty-one cases of fully developed dystrophia myotonica, fifteen showed changes in the skull. Thicken-
ing of the calvaria and hyperostosis interna were present in eleven cases, large frontal sinuses in eight, a small to very small sella turcica in thirteen, prognathism in three, and calcification of the falx in three. Jequier (1950), and de Oliveira and Mosinger (1951) also discussed the skull changes in dystrophia myotonica. Walton and Warrick (1954) studied seventeen cases and confirmed the high incidence of thickening of the calvaria and the small pituitary fossa. In none of their patients did they find hyperostosis interna and they did not consider enlargement of the frontal sinuses, bridging of the sella, prognathism, or intracranial calcification as being statistically significant.

The object of the present publication is a survey of the skull roentgen films of eighteen cases of dystrophia myotonica which have been investigated at the National Institute of Neurological Diseases and Blindness in the past five years. The records of these patients have been reviewed and the diagnosis of dystrophia myotonica is beyond doubt. Sixteen of the eighteen patients included in the present study had muscle biopsy. Usually the diagnosis of dystrophia myotonica is made on clinical basis: muscle biopsy however may yield strong confirmatory evidence.

Our findings are summarized in the Table on p. 23. Sixteen out of eighteen cases (89 per cent) showed changes of the vault. Such changes which we will indicate for the matter of simplicity with the general term of 'hyperostotic' may be grouped in four different types:

1. Thickened calvaria throughout — nine cases (Fig. 1). This type of change corresponds to what Moore (1955) indicated as 'hyperostosis calvariae diffusa'.
2. Thickened calvaria in the frontal region — two cases (Fig. 2), corresponding to the 'nebula frontalis' of Moore.

3. Hyperostosis frontalis interna — three cases (Fig. 3). In the frontal region, irregular thick mammillated bulgings of newly formed bone project towards the endocranium, and an unaffected 'groove' along the midline is clearly seen. This type of change corresponds to the 'hyperostosis frontalis interna' of Moore. However, our criteria for making the diagnosis of hyperostosis frontalis interna are somewhat different. The 'nodular' appearance of the inner surface of the frontal bone as well as the unaffected midline 'groove' are in our opinion important roentgenographic features of this condition.

4. Thickened calvaria throughout and hyperostosis frontalis interna — in two cases (Fig. 4).

These 'hyperostotic' changes of the vault occurred with the same frequency in males and females (eight males and eight females) which is a striking fact when we consider the sex incidence of the large series of cases of hyperostosis cranii reported by Henschel (1949), 99 per cent in women, and by Moore (1955), 98.2 per cent in women. The age distribution in our series was as follows: second decade, two; third decade, three; fourth decade, seven; and fifth decade, six cases. All patients in the fourth and fifth decades had changes. The two cases without 'hyperostosis' were aged eleven and twenty-two. The more severe cases occurred in women and the longer the duration of the disease,
as gauged by the duration of the patients' symptoms, the more advanced were the 'hyperostotic' changes.

An attempt was made to correlate the hyperostosis cranii with the onset of gonadal atrophy. In four males we had clinical evidence of gonadal atrophy which was confirmed by gonadal biopsy. In one other male the testicles were very small. The assessment of ovarian hypofunctions is more difficult. Two of our female patients, both in their fourth decade, showed a high level of follicle stimulating hormone (greater than 172 mouse units in one and greater than 96 mouse units in the other). In another woman hysterectomy and oophorectomy had been carried out at thirty-eight years of age. In these eight patients who had evidence of gonadal failure, the degree of hyperostotic changes was most marked.

In three of the eighteen cases the shape of the skull was acromegaloïd. In seven cases the frontal sinuses were very large, large, or moderately large; in two cases a large sphenoidal sinus was present. No significant elongation of the mandible was found in the present material. The 'small size of the sella' and the intracranial calcifications reported in other publications are not by themselves findings of proven pathological significance.

Concerning possible etiological factors accounting for the 'hyperostotic' changes in patients with dystrophia myotonica, one of us in a previous publication (Caughey 1958) has suggested that hyperostosis cranii, excessive sinus formation, and prognathism are acromegaloïd features brought about by
unrestrained activity of growth hormone, released as a result of the gonadal failure. The possible relationship of the hypogonadal function and the degree of skull changes reported in our present series, lends support to this contention. Another possibility is that the skull changes may be on a genetic basis, as a pleiotropic manifestation of a single gene or an expression of involvement of different genes.

Further studies of the problem could include:

1. Radiologic examinations of the skulls of relatives of affected patients.

2. Periodical roentgen examinations of patients with dystrophia myotonica to establish possible progression in the hyperostotic process. Two of the patients of the present series have been roentgen examined on two occasions. The interval between the examinations was two years in one and eighteen months in the second case. After this lapse of time no significant difference was seen in the hyperostotic changes which were present in both of these patients. Muscle lesion had progressed in both cases during the interval between the two admissions.

3. Search for evidence of excessive growth hormone when such measurement becomes available.

SUMMARY

The skull roentgen films of eighteen cases of dystrophia myotonica have been reviewed. Seventeen had 'hyperostotic' changes of the vault, the sex distribution being equal. The longer the duration of the disease the more marked were these changes. In eight cases with proof of hypogonadism the hyperostosis was most advanced. Because of their frequency hyperostotic changes should be considered as one of the variable features of the disease.

ZUSAMMENFASSUNG


RÉSUMÉ

Les auteurs ont examiné les radiographies du crâne de dix-huit cas de dystrophie myotonique. Dans dix-sept cas il y avait des modifications 'hyperostotiques' de la voûte, également distribuées dans les deux sexes. Les modifications hyperostotiques étaient d'autant plus marquées que la durée de la maladie avait été plus longue. Dans huit cas présentant un hypogonadisme prouvé, l'hyperostose était très marquée. En raison de leur fréquence, les modifications hyperostotiques devraient être considérées comme un des signes variables de cette affection.
REFERENCES


