Clinical and radiographic dental findings in X linked hypohidrotic ectodermal dysplasia

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Abstract

X linked hypohidrotic ectodermal dysplasia was studied in the dentition of both affected males and carrier females. Hypodontia was more severe in males than females and there were differences in the pattern of tooth absence between the sexes. Abnormal crown form, with the maximum diameter of the teeth being apically displaced, was noted particularly in the anterior teeth. Taurodontism was commonly seen radiographically.

X linked hypohidrotic ectodermal dysplasia (XHED) is expressed fully in males and partially in heterozygous female carriers. It is characterised by sparse hair, diminished sweating, and dental anomalies, particularly hypodontia (congenitally missing teeth). The prevalence in the population has been assessed as lying between 1:10 000 and 1:100 000 male live births.¹

There are a number of reports of the dental findings in individual cases and their families, together with four substantial investigations.¹⁻⁴ Nakata *et al*² determined tooth size in 49 subjects in 15 families with XHED. Affected males and carrier females both showed consistently smaller mesiodistal tooth crown diameters than did unaffected persons. Radiographs were taken but were not reported on separately. Airene³ studied 11 males from nine families together with their relatives. He confirmed the reduction in mesiodistal diameter and showed that this was also true for the buccolingual dimension in both affected males and heterozygous females. Radiographs were used only to assess the degree of hypodontia. Soderholm and Kaitila⁴ investigated six patients and their five

mothers, the heterozygous mothers having hypodontia affecting four or more teeth. A detailed analysis of the dental anomalies does not seem to have been published. Radiographs were not reported to have been taken routinely. Clarke¹ examined 35 families with XHED. He found the mean number of deciduous teeth absent in affected males to be 16.4 (out of the normal 20) and the mean number of permanent teeth absent to be 23.7 (out of the normal 28, excluding third molars). Seventy-five percent of obligate carrier females had 'distinctly abnormal' permanent teeth. Radiographs were only occasionally available.

It was the aim of this study to assess the dental findings in affected males and heterozygous female members of families with XHED using 'distant examination' techniques.

Subjects, materials, and methods

Thirty-four British families with XHED were identified as part of a clinical and genetic study of this disorder.^{1,5} Both males and females from these families had previously been examined by one of the present authors (AC). The District Dental Officer for each of these subjects was subsequently asked for cooperation in arranging a clinical and radiographic dental examination. Forty of the 42 approached kindly agreed. Patients were then contacted by letter explaining the nature and purpose of the investigation and enquiring of them their willingness to participate and the name of their present dental surgeon (if any). Letters were sent to 140 persons and 62 affirmative replies were received.

Either the patient's own dental surgeon or the appropriate Community Dental Service was supplied with an explanatory letter, a copy of the patient's signed consent form, details of the ethical approval, and an examination form. It was intended that, wherever possible, the special examination would form part of the patient's routine recall and the radiographs would be available for return to the practitioner. Practitioners were asked to allocate teeth examined clinically to either 'normal', 'conical/peg', or 'reduced' categories on the basis of crown form and size. For convenience, the last two groupings were subsequently combined by us to give a single 'abnormal' code for data analysis. The completed examination

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forms and radiographs were returned by post to the authors.

The radiographs were viewed by the first two authors under standardised conditions in conjunction with the clinical record until a consensus view was reached regarding the various parameters to be recorded (fig 1). Teeth were deemed 'present' if at least one of any tooth type was present, or if there was radiographic evidence of tooth removal, or if the general practitioner had recorded the tooth as having been previously extracted. Teeth were only classed as 'absent' if none of these criteria was met. The authors also assessed the position of the maximum mesiodistal convexity of the tooth crown, corresponding to the contact area between teeth. It was felt that if this was situated in the apical half of the crown, this feature would correspond with 'conical' anterior tooth form and 'bud-shaped'³ molar crown form. Despite

repeated efforts at quantitation of taurodontism, both by the present authors and others, no single biometric method has been universally accepted.^{6 7} For the purposes of the present study, the authors found themselves to be consistently in agreement using a subjective assessment. One quarter of the radiographs were considered randomly again under identical conditions with no significant disagreement from the first viewing.

Results

From the postal and radiographic study, usable records were available for 22 affected males with a mean age of 19.4 years (median 11 years, range 3 to 65 years) and 23 carrier females with a mean age of 28 years (median 28 years, range 8 to 76 years). Affected males and carrier females showed differences in the

Figure 1 (Top) Orthopantomogram of affected male showing severe hypodontia, conical incisors, bud shaped molars, and taurodontism. The contact points of all teeth are apically displaced. (Bottom) Orthopantomogram of manifesting carrier female showing hypodontia, tapering incisors, bud shaped molars, and taurodontism. The contact points of all teeth are apically displaced to a lesser degree than in the male.

absence of teeth, both in terms of number and tooth type affected (fig 2A). Only the males were of an age to make an assessment of deciduous tooth hypodontia worthwhile. The pattern of absence is illustrated in fig 2B. In the permanent teeth of affected males, a clinically abnormal crown form was observed more frequently in the anterior teeth than in the molar region. In carrier females, there was a slight increase in prevalence of clinically abnormal crown form from the midline to the canine region with a subsequent

trend towards normality more posteriorly (fig 2C). The vast majority of males showed apical displacement of the contact points in all three groups of teeth. In females the prevalence of this abnormality was less, with a greater tendency for these abnormal crown forms to be found anteriorly (fig 2D). The findings were similar for deciduous teeth, subject to the limitations imposed by small numbers, particularly in the females (fig 2E). Almost all male first and second molars were categorised as taurodont. Female molar

Figure 2 Prevalence of dental abnormalities in XHED males and females. (A) permanent tooth absence, (B) deciduous tooth absence (males only), (C) abnormal crown form in the permanent dentition, (D) apically displaced contacts in the permanent dentition, (E) apically displaced contacts in the deciduous dentition, (F) taurodontism. Deciduous teeth are referred to as A and B (the incisors, I), C (the canines), D and E (the molars, M). Permanent teeth are referred to by number from the midline, anterior to posterior, 1 and 2 (the incisors, I), 3 (the canines, C), 4 and 5 (the premolars, PM), 6, 7, and 8 (the molars, M).

teeth were less often affected, with the prevalence of taurodontism falling from first to third molars (fig 2F).

The radiographs showed a number of additional findings previously unreported in association with XHED. These included a supernumerary lower incisor tooth in a female; two females and one male with 'talon cusps' (an evagination of the tooth crown, usually on the palatal aspect of an incisor or canine tooth); 'shovel shaped' upper central incisors (a finding most commonly seen in mongoloid populations) in two females; one case of 'gigantiform', almost twice the expected size, third molars. One family also showed a reverse curvature to the occlusal plane.

Although there seemed to be an almost equal proportion of males and females affected by asymmetry of tooth number, there was evidence of greater disturbance of tooth form in females than males. The most notable example of this was one female case with a marked delay in the formation of teeth in one quadrant. Further investigations into the three dimensional symmetry of tooth crowns and the detailed radiographic findings in these cases are under way.

Discussion

PERMANENT TOOTH HYPODONTIA

Brook⁸ stated the prevalence of permanent tooth hypodontia to be 5.7% in females and 3.1% in males aged 11 to 14 years. Silverman and Ackerman⁹ showed that, where there was hypodontia, 80% of affected subjects had one or two missing teeth and only 5% had five or more missing teeth. The present findings of a mean of 23.7 missing teeth in affected males recorded using 'distant examination techniques' is almost identical to the previous clinical findings of one of the authors.¹ It was interesting to note that there was a substantial inter- and intrafamilial variation in the number of teeth absent, the latter varying from 0 to 28 in the same pedigree.¹ The mean number of missing teeth in carrier females in the present study was four; 15% had no missing teeth, 20% had one or two missing, and 35% of the sample had five or more teeth absent. These proportions are markedly different from those reported for the general population.^{8,9}

The pattern of hypodontia seen in the affected males resembles that reported by Nakata *et al*² and Tso *et al*¹⁰ in that the first molars, upper central incisors, and upper and lower canines were the most 'stable' tooth types, that is, they were most often present. Both the assessment of hypodontia¹¹ and of hypodontia with unusual tooth morphology⁵ have been used to investigate females in affected families. In both of these studies, hypodontia has shown a correlation with the results of sweat tests and may thus provide a simple screening test for the carrier status. From the present findings it is evident that

dental radiographs can provide useful additional information in cases of uncertain diagnosis. A further advantage is that such material may be reviewed in the absence of the patient by an experienced clinician.

Hypodontia in carrier females in the present study was different from that in males firstly in severity and secondly in the pattern of tooth absence, resembling that reported by Schulze¹² in severe, non-specific hypodontia. This was most noticeable in the case of first premolars which were relatively protected in females but almost universally absent in males. This finding was mirrored by the frequent absence of deciduous predecessors of the first premolars in the males. We do not know whether the deciduous second molar is correspondingly absent in females, but intend to conduct a longitudinal study to clarify this point.

DECIDUOUS TOOTH HYPODONTIA

On examination the mean number of teeth in the deciduous dentition of 41 affected males was 3.6. The teeth were typically peg shaped or conical and abnormal molar crown forms were also noted.¹ Both the clinical and radiographic examination showed a relative 'protection' of deciduous canines and second molars in males, contrasting with the high rate of hypodontia in the upper deciduous incisors and mirroring the findings in the permanent dentition.

The deciduous teeth of three obligate carriers of XHED were examined in childhood.¹ Two 5 year old girls had spacing resulting from microdontia especially affecting the incisors and canines. One 8 year old girl had no maxillary central incisors and the mandibular central incisors were peg shaped. Seven of the 16 adult obligate carrier females who could clearly remember their deciduous teeth described them as having been abnormal, with missing, peg shaped, or small teeth. In total, the deciduous teeth were abnormal in 10 out of 19 obligate carriers. Brook⁸ has described hypodontia in the female deciduous dentition as being very rare (0.1 to 0.9% in 20 000 children) and Sofaer¹¹ suggested that this would be a useful feature for the screening of females for possible carrier status for XHED. In this study, insufficient females were examined in the deciduous or mixed dentition stages to allow comment on this proposal.

CROWN FORM

The heterozygous females showed a tendency for the more abnormal crown forms to be present in the anterior teeth. It remains to be seen whether this finding is an expression of the carrier state of XHED or whether it is a more general feature of the female dentition. It may be that the clinical assessment of altered crown form is more easily made anteriorly than posteriorly.

TAURODONTISM

The definition of taurodontism was perhaps most clearly given by Stenvick *et al*⁷ as a "larger than normal body to the tooth". This usually corresponds to an apical migration of the furcation of the roots, together with loss of the cervical constriction of the tooth. The pulp form tends to follow the root form, resulting in long, wide, straight sided pulp chambers. Although the condition is most frequently recognised in molar teeth, corresponding findings are present in single rooted teeth.^{13 14} There have been frequent allusions to large pulps being a feature of XHED but an authoritative source reference is apparently lacking. Our findings indicate that taurodontism is a regularly occurring feature of XHED.

Stoy¹⁵ reported taurodontism in association with sparse hair and hypodontia in a 10 year old female but no mention was made of the patient's general condition. This has been termed the 'sparse hair, oligodontia, and taurodontism' (SOT) syndrome and may be XHED by another name.

THE VALUE OF DENTAL RADIOGRAPHS IN THE DIAGNOSIS OF XHED

In previous investigations of XHED families,⁵ possible carrier females have been identified by means of pedigree analysis and hypodontia. Of the families in the present study, the carrier status of some females could not be confirmed because of difficulty in ascertaining the past dental history. Without accurate information regarding past extractions, it is impossible to be certain which teeth were originally congenitally absent. Radiographic examination is particularly useful in the mixed dentition when the presence or absence of permanent teeth as yet unerupted may be noted.

Of the obligate carrier females studied, three out of 14 had a full complement of permanent teeth. By study of the crown and root forms as described, it was possible to augment the clinical assessment of these subjects, since all who had teeth showed morphological features characteristic of their carrier status. In one case of an adult carrier female the deciduous canine teeth were retained. The radiograph showed the presence of the unerupted permanent successors impacted in the palate. This is not uncommon in the normal population and the permanent canines have been shown in this study to be rarely absent in XHED females. This highlights the need for the clinician to

be familiar with the pattern of tooth absence in this condition and to have supporting evidence to hand, such as radiographs or linkage analysis, before discussing recurrence risk with individual patients. An additional 35 adult obligate carrier females have been studied clinically.¹ Ten of these had apparently normal dentition; four had two missing teeth (of whom three had further dental features of XHED), 15 had more than two absent teeth, and six had other dental anomalies such as microdontia or conical teeth or both without hypodontia. Dental radiography will be of value in the investigation of the first of these groups to clarify their possible carrier status.

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