The Trismus-Pseudocampylodactyly Syndrome

B. G. A. TER HAAR* and R. F. VAN HOOF†

Summary. A new family with the combination of trismus and curvature of the fingers on dorsiflexion of the wrist is described. Data from 24 affected members of this family make it likely that the measurements of mouth opening and of wrist angles are sufficient to recognize the syndrome. The affected persons also show a mild degree of short stature but only when compared with their unaffected sibs of the same sex. In the pronounced cases of trismus the coronoid process is enlarged by the extensive pull of the temporal muscle tendon unit which decreases the mandibular excursion. Surgical correction of the enlarged coronoid process may then be necessary.

The variable expressivity of this autosomal dominant trait makes it most likely that the phenotypes in the families described hitherto are due to the same mutation.

Simultaneously in 1969 Hecht and Beals, and Wilson et al published their findings on a new syndrome. This consisted of trismus, campylodactyly, feet deformities, and a shorter than average stature.

Hecht and Beals described a family with an affected father, normal mother, three affected children (two boys and a girl), and three normal children. One other girl died at 13 days of age. The paternal grandmother was questionably affected. A similar combination of defects in another family was noted by Wilson et al (1969). Thirty-eight family members were examined through three generations and 14 were found to have varying degrees of involvement. The names and origins of these pedigrees were compared but no common family names or origins were noted. Both authors had knowledge of a third family in Saskatchewan. Data on this were presented by Surana and Ives (1971) and it was shown that they had emigrated in 1913 from the Netherlands and a comparison of family names makes it most likely that this is the same family as the one reported in the present study.

Discussing the mechanism of limitation of excursion of the lower jaw, Surana and Ives (1971) stated that no radiographic abnormalities of the temporomandibular joint had been identified and that the basis of this strikingly unusual finding was not known. In search for an explanation of the trismus in our patients we also found no abnormalities in the temporomandibular joint, but were able to confirm an enlargement of the coronoid process. This bony abnormality encroaches upon the available space in the infra-temporal fossa. The enlarged coronoid process impinges upon the body of the zygomatic bone and inner margin of the arch which explains the limited mandibular excursion (Van Hoof, 1973).

Hecht and Beals (1969) considered the trismus as the predominant feature, whereas Wilson et al (1969) considered the campylodactyly as the major symptom. Hecht and Beals tended to regard the phenotype of Wilson's family as stemming from a different mutation and also noted a shorter stature than average in their patients.

The purpose of our family investigation was to delineate the syndrome by measuring hand movements, mouth openings, and body length in order to test the idea that all the patients described in the literature and our family can be regarded as the same phenotype.

Clinical Features of the Trismus-Pseudocampylodactyly Syndrome

The syndrome includes:

1. Trismus. Due to restricted mouth opening the patients present mastication problems and may

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need twice the usual time to consume an ordinary meal. Besides this the affected individuals may reveal all distressing symptoms of trismus, such as lack of hygienic dental care because normal dental treatment is very difficult to carry out. Moreover intubation in case of any surgical procedure is troublesome. Severe trismus may give rise to a life threatening situation as patients have been known to aspirate vomit very easily.

The mouth opening is measured in millimetres between the incisal edges of the central upper and lower incisors. To classify a mouth opening as restricted one has to know normal values. Kusen (1960) and Derksen (1970) investigated these values in adults as listed in Table I, and Kusen also measured the mouth opening in 530 schoolchildren ranging from 7 to 18 years and he did not find a significant difference between boys and girls (Table II).

Measuring between the alveolar ridges is also unreliable because the differences in resorption cannot be taken into account.

2. Pseudocampylodactyly. Dorsiflexion of the wrist results in a curvature of the fingers (except the thumb), which occurs at all the interphalangeal joints. Volar flexion of the wrist allows complete extension of the fingers. There is no muscle weakness detectable. Radiography of the hand and forearm never revealed any bony abnormalities of the radius or ulnar nor of the bones of the hand. The hand deformities are much like those described by Smith and Kaplan (1968) in their extensive review on campylodactyly, but there are several reasons which make us prefer the name pseudocampylodactyly: (1) the lack of progression, (2) the absence of fixed deformities, (3) the involvement of the flexor sublimis muscle as well as the flexor profundus, and (4) the presence, in pronounced cases, of ulnar deviation and a mild limitation of dorsiflexion and supination of the hand so one can presume that there is involvement of the flexor muscle tendon units of the wrist as well.

Functional disability from the pseudocampylodactyly is not usually great and therefore most patients do not wish surgical treatment. The only time the deformity was a significant handicap occurred in a man during his army base training when his drill instructor criticized him for doing press ups on his knuckles (Wilson et al., 1969). One of our patients also had problems while on military service because he was unable to give the correct salute but he was given special dispensation.

In several affected subjects, a history of tightly clenched fists at birth was volunteered. Subsequently this usually disappeared after exercises carried out by the mother. During childhood an interesting finding may occur since, because of the deformity of the hands, the affected child can only crawl on his knuckles and this may give rise to the formation of a callosity.

In recording the pseudocampylodactyly the maximal active extension in the wrist with stretched fingers is measured. The method of measuring and recording is based on the principles of the Neutro-Zero Method (Cave and Roberts, 1936). The extended 'anatomical position' of the wrist is therefore accepted as zero degrees. According to the American Academy of Orthopaedic Surgeons (1965) the average extension of the wrist is $71^\circ$. They quoted this as an overall average of several investigations ranging from $60^\circ$ to $90^\circ$.

3. Foot Deformities. In the trismus-pseudo-
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4. Shorter than Average Stature. To evaluate any growth retardation, the affected individuals should be compared with their unaffected sibs of the same sex as well as with the 10th-centile measurements of the population.

All affected individuals in our family have normal intelligence. Wilson et al (1969) recorded two patients who also had mild prognathism. We noted this in four of our patients, but this is considered a coincidence. In two patients bilateral dislocation of the hip was revealed, but because they are in the same sibship this is also considered to be a coincidence.
Family Investigations

Our family investigation started with two propositi, and resulted in the pedigree shown in Fig. 1.

Propositus 1 (VI.6) was referred to the oral surgery department for evaluation of his trismus. The trismus was of unknown duration and had not given any serious problems until it interfered with adequate dental treatment. The incisal clearance was 13 mm. Radiology showed a normal joint but a bilateral vertical elongated coronoid process (Fig. 2). By means of a mouth dilator some elastic resistance was felt, so exercises were started. The opening has been stationary at 20 mm for almost four years. Coronoidecetomy was planned at the end of puberty but the patient refused to be operated upon since he had no functional problems and dental care could be carried out with an acceptable difficulty.

Propositus 2 (VI.61) was referred to the paediatric department because of multiple congenital deformities by bilateral dislocation of the hip, clubfeet, ulnar deviation of the hands, and flexed fingers on dorsiflexion of the wrist but normal extension on volar flexion (Fig. 3). In this complicated case history the symptom of trismus was not noted, but the parents drew our attention to it. The mouth opening was found to be 10 mm.

Because of the common family name, a rather rare one, of propositi 1 and 2 connection between the two patients was suspected. To establish the family relationship was a difficult investigation, as it was necessary to go back in history for five generations until the pedigree yielded common ancestors at the end of the 18th century. We tried to examine most of the living members personally but sometimes access to the homes was not easy because the religious beliefs of these people made them regard congenital deformities as a punishment of God, and they are therefore ashamed of them.

The case histories of two patients are considered to be of special interest and are therefore described in more detail.

VI.8. This patient is the brother of propositus 1, and was first seen at the clinic at the age of 20, some months after his brother's first examination. He had had trismus since childhood and at 3 years could only open his mouth 3 mm. The causative factor was an enlarged coronoid process which was operated upon in another hospital four times (because of three recurrences). The few teeth left were decayed. Radiology and laminograms showed bilateral coronoid process enlargement comparable to that of his brother but it was surrounded by a considerable mass of osseous callus, probably induced by the previous operations. The confines of the sigmoid notch had almost disappeared. Therefore bilateral horizontal osteotomy of the vertical ramus was performed by an extra-oral submandibular approach. After this operation intra-oral splints were inserted in order to prevent dislocation of the mandibular body. With the help of these splints and elastic bands elaborate exercises were performed. This prevented both the recurrence of the condition and dislocation of the mandibular body.

Opening of the mouth to 32 mm is now possible. The follow up revealed no recurrence three years after the operation. This patient also has moderate pseudocampylodactyly which gives no functional disability.

V.20. This patient is the father of propositus 2. Since childhood there had been moderate trismus, which had not troubled the patient much but dental treatment had been difficult. Pseudocampylodactyly was pronounced. He had no functional disability but 12 years ago he was operated upon by a surgeon who performed an operation to improve the function of his left hand. He had hoped to operate successfully upon the patient's son who also showed severe pseudocampylodactyly. Therefore the father volunteered to have his left hand operated
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Fig. 3. Pronounced case of pseudocampylodactyly in propositus 2 (VL61) showing ulnar deviation of the hands both clinically and radiologically (top and centre).

Because of the pseudocampylodactyly the child can only crawl on his knuckles (lower right), which gives rise to the formation of a callosity (lower left).
TABLE III
PATIENTS WITH THE TRISMUS-PSEUDOCAMPYLODACTYLY SYNDROME (TPS) AND THEIR SIBS (N)

<table>
<thead>
<tr>
<th>Case</th>
<th>Sex</th>
<th>Right</th>
<th>Left</th>
<th>Average</th>
<th>Maximal Active Extension in the Wrist with Stretched Fingers</th>
<th>Mouth Opening* (mm)</th>
<th>Length (cm)</th>
<th>Age (yr)</th>
</tr>
</thead>
<tbody>
<tr>
<td>V.3</td>
<td>N</td>
<td>+70</td>
<td>+75</td>
<td>+72.5</td>
<td>43(p)</td>
<td>163</td>
<td>53</td>
<td></td>
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<tr>
<td>V.4</td>
<td>TPS</td>
<td>-15</td>
<td>0</td>
<td>-7.5</td>
<td>42(p)</td>
<td>159</td>
<td>52</td>
<td></td>
</tr>
<tr>
<td>V.5</td>
<td>TPS</td>
<td>+30</td>
<td>+35</td>
<td>+32.5</td>
<td>40(p)</td>
<td>161</td>
<td>52</td>
<td></td>
</tr>
<tr>
<td>V.6</td>
<td>TPS</td>
<td>-10</td>
<td>-20</td>
<td>-15</td>
<td>47(p)</td>
<td>159</td>
<td>51</td>
<td></td>
</tr>
<tr>
<td>V.7</td>
<td>TPS</td>
<td>0</td>
<td>-10</td>
<td>-5</td>
<td>21</td>
<td>171</td>
<td>47</td>
<td></td>
</tr>
<tr>
<td>V.8</td>
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<td>+25</td>
<td>+15</td>
<td>+20</td>
<td>32</td>
<td>161</td>
<td>44</td>
<td></td>
</tr>
<tr>
<td>V.9</td>
<td>N</td>
<td>+60</td>
<td>+70</td>
<td>+65</td>
<td>49</td>
<td>163</td>
<td>42</td>
<td></td>
</tr>
<tr>
<td>V.11</td>
<td>N</td>
<td>+55</td>
<td>+65</td>
<td>+60</td>
<td>47(p)</td>
<td>170</td>
<td>69</td>
<td></td>
</tr>
<tr>
<td>V.12</td>
<td>N</td>
<td>+60</td>
<td>+65</td>
<td>+65</td>
<td>44(p)</td>
<td>158</td>
<td>66</td>
<td></td>
</tr>
<tr>
<td>V.13</td>
<td>N</td>
<td>+45</td>
<td>+35</td>
<td>+40</td>
<td>36</td>
<td>176</td>
<td>65</td>
<td></td>
</tr>
<tr>
<td>V.14</td>
<td>TPS</td>
<td>+5</td>
<td>+20</td>
<td>+12.5</td>
<td>9</td>
<td>155</td>
<td>64</td>
<td></td>
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<tr>
<td>V.15</td>
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<td>35</td>
<td>165</td>
<td>62</td>
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<tr>
<td>V.17</td>
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<td>+65</td>
<td>46</td>
<td>167</td>
<td>57</td>
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<tr>
<td>V.18</td>
<td>TPS</td>
<td>+20</td>
<td>+35</td>
<td>+27.5</td>
<td>28(p)</td>
<td>165</td>
<td>55</td>
<td></td>
</tr>
<tr>
<td>V.19</td>
<td>N</td>
<td>+55</td>
<td>+65</td>
<td>+60</td>
<td>45</td>
<td>158</td>
<td>53</td>
<td></td>
</tr>
<tr>
<td>V.20</td>
<td>TPS</td>
<td>-50</td>
<td>(+10)</td>
<td></td>
<td>31</td>
<td>171</td>
<td>51</td>
<td></td>
</tr>
</tbody>
</table>

The patients who were operated upon for pseudocampyloectomy are given in parentheses.
* (p) stands for prosthesis; the values in these patients are taken between the upper and lower incisors as well.

Fig. 4. Case V.20 who is unable to extend the fingers at the interphalangeal joint, when the wrist is in dorsiflexion. The patient’s left hand was operated upon successfully.

Results

Measurements. The results of the measurements in this family are given in Table III and there appeared to be no significant difference whether the patient was right- or left-handed. In Fig. 5 the correlation between the mouth opening and the wrist angles at which bending of the fingers started is shown. The population of the ‘normal’ family members is clustered and is characterized by on, thus giving the surgeon the opportunity to get acquainted with the technique required to handle this rare condition. As can be seen from Fig. 4, the operation was a success. Afterwards the patient’s son (VI.58) was also operated on successfully in both hands.
to diagnose a coronoid process enlargement. Only the affected children (VI.6, VI.7, VI.8) showed abnormalities, whereas the mother (V.4) and unaffected children (VI.5) had a normally defined coronoid process. Eight of the total number of 24 affected individuals were x-rayed and all exhibited coronoid enlargement. In several of the more pronounced cases, the sigmoid notch was small, as there was a backward curvature of the posterior border of the coronoid process. The clinical and radiological features of father (V.20) and his offspring may illustrate a correlation between the seriousness of the trismus and the extent of the coronoid enlargement (Fig. 6). VI.60, who has a moderate restriction of mouth opening, seems to have only a slightly enlarged coronoid process.

**Genetics.** A dominant pattern seems likely as occurrence in successive generations with transmission to half of the offspring on average was noted while the unaffected individuals did not give the syndrome to their children. Male to male transmission was noted several times, which indicates that transmission occurs by one of the autosomes.

**Discussion**

Measuring the mouth opening and wrist angles offers sufficient data to divide the family members into separate groups of affected and unaffected persons. Using only one of the parameters is insufficient because of the extremes. The lack of a direct relation between the trismus and restricted wrist angle, also indicates the variable expressivity of the syndrome. It is not possible to observe differences between the sibships. The noted variable expressivity in this family strongly suggests that the families investigated by Hecht and Beals (1969) and Wilson et al. (1969), can be regarded as stemming from alleles arising by mutation of the same gene.

The body length in affected individuals is not under the 10th centile for the Dutch population, but is less than the height of unaffected sibs of the same sex. Hecht and Beals (1969) suggested that this mild shortness of stature may result from short muscles preventing the normal growth of the epiphyseal plates. It seems likely that more muscles are involved than those of fingers, toes, and jaw.

The coronoid process enlargement offers a good explanation for the trismus in the pronounced cases. Considering the normal relation of the coronoid process and the temporal muscle tendon unit, and especially the almost horizontal fibres which pull the jaw backwards, one can draw the following conclusion: the unusual shape of the coronoid process

mouth openings of more than 35 mm and wrist angles of more than 40°. The patients show much lower values but they are occasionally not far beyond these limits. There are two exceptions: VII.24 has a normal mouth opening but a restricted wrist angle; VII.12 with a rather normal wrist angle, has a small mouth opening.

If the relation between trismus and restricted wrist angle was a direct one, all patients should have been arranged along a line, but considering Fig. 5, this is not demonstrated.

The children of IV.1 include two unaffected females both with a height of 163 cm, while the affected females have an average height of 160 cm (159, 161, 159, and 161 cm). Two male offspring of IV.2 are affected: average height 168 cm (165 and 171 cm) and one is unaffected: height 170 cm. As with the children of IV.1, so for those of IV.2 where the affected offspring (V.14) is shorter (155 cm) than her unaffected sibs (average height 164-8 cm: 158, 176, 165, 167, and 158 cm). The two sisters VI.5 and VI.7 can also be compared: the affected girl measures 161 cm and her unaffected sister has a height of 175 cm. No other sibs could be compared since they were too young and their ages too scattered.

**Radiological Findings.** Radiological investigation in V.4 and all her children was made in order
Fig. 6. Top: father (V.20) with moderate trismus and coronoid process enlargement (arrows). Note the backward curve of the process leaving a small sigmoid notch.
Centre: his son (VI.61) with severe trismus and coronoid enlargement (arrows).
Bottom: his daughter (VI.60) with about 5 mm mouth opening beyond the average has only a slightly enlarged process (arrows).
in this syndrome confirms the hypothesis that coronoid enlargement in these cases is caused by extensive pull by the temporal muscle tendon unit.

The mode of inheritance of the syndrome is by an autosomal dominant gene with full penetrance and variable expressivity without sex influence.

The combination of hand deformities and trismus is also found in the whistling-face syndrome and in a syndrome which consists of hemifacial microsomia and aplasia of the radius (Ter Haar and Van Hoof, 1973). Furthermore, O'Rourk and Bravos (1969) described a boy with a small oral orifice and hand abnormalities and they proposed the condition should be known as the oculo-dento-digital syndrome. In doubtful cases these other syndromes have to be considered.

References

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