Case report

The patient, a 9.3 year-old girl, was delivered by cesarean section, because of breech presentation, after a full-term, uncomplicated pregnancy, with no exposure to radiation or any unusual agent. Birth length (51 cm), weight (3.600 gr) and head circumference (33 cm) were normal. The APGAR score was 6. As a neonate she was hospitalized for failure to grow and gain weight. She had bilateral hip and developmental milestones were retarded. At 3.5 months she was in an infant incubator for 2 days for respiratory distress. Her growth was 154+4.5 cm. Peak growth values post Arginine and glucagon were 5.78 and 12 ng/ml, respectively (normal>10). Delayed eruption of a complete, but with single maxillary central incisor, primary dentition was reported. No consanguinity and similar case in the family was mentioned. The target height was 154+4.5 cm.

On physical examination at age 9.3 years her height was 117 cm (SDs -3.1), and her weight 21 Kg (< 5th centile). She had microcephaly (head circ.: 50 cm), brachycephaly (ceph. index: 83.8), groove along the coronal suture, low set and prominent ears, hypotelorism, short and narrow upper face, hypoplastic maxilla, narrow, mixed dentition with single maxillary permanent central incisor (Figure 1) highly arched palate (Figure 2), and hyperextensibility of joints. She demonstrated a borderline mentality. Measurements on casts (age 10.6) showed a short premaxilla (1.1 cm), narrow palate (palatal width 2.5 cm, -2.5 S.D.), high palate (palatal height 1.75 cm, >2S.D.)?. A panoramic radiograph (done at age 8) showed hypodontia of the permanent dentition; the maxillary lateral and the four mandibular incisors were congenitally missing (Figure 3).

Cephalometric analysis (Table 1) revealed severely decreased anterior and posterior cranial base, severely increased cranial base saddle angle (platybasia), severely tilted up anterior cranial base, severely short anterior and posterior upper facial height, severely short maxilla, severely retruded maxilla, concave profile, severely retruded maxillary denture base, well positioned mandible relatively to the maxilla, severely retruded maxilla, narrow, mixed dentition with single maxillary permanent central incisor (Figure 1) highly arched palate (Figure 2), and hyperextensibility of joints. She demonstrated a borderline mentality. Measurements on casts (age 10.6) showed a short premaxilla (1.1 cm), narrow palate (palatal width 2.5 cm, -2.5 S.D.), high palate (palatal height 1.75 cm, >2S.D.)?. A panoramic radiograph (done at age 8) showed hypodontia of the permanent dentition; the maxillary lateral and the four mandibular incisors were congenitally missing (Figure 3).

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in the median structures of the face is more severe in our case than in the reported so far in the literature, since none of those had this hypodontia in addition to the single maxillary central incisor. The girl also had microcephaly, severe hypotelorism (interpapillary distance 44 mm, sufficiently below the 3rd percentile), and borderline mentality. Although smell was reported to be functional and a CT brain scan at the age of 18 months disclosed no brain dysmorphology, the essential question of a minor defect of the midline structures of the forebrain not detected by the scanning still remains. It has been emphasized that SMCI may occur alone or in other conditions bearing no relationship to holoprosencephaly. Nevertheless, the pathogenetic similarity of the SMCI and short stature syndrome to holoprosencephalic microforms is obvious, although no etiologic relationship has been established. However, individuals with SMCI with or without hypotelorism, or microcephaly in families with A.D. form of holoprosencephaly had children with holoprosencephaly. On the other hand, all the reported cases with the syndrome of SMCI and short stature were sporadic, all but one female who didn’t have any pregnancies, had not reached reproductive age and there is no follow-up information about their reproduction. Nothing is known about the genetics and the recurrence risk of the syndrome. So the risk for individuals with the syndrome of having holoprosencephalic children is not known.

In order to give answers to these questions and more information on the phenotype of the syndrome we did cephalometric analysis

Table 1. Cephalometric analysis for case report

<table>
<thead>
<tr>
<th>Measurement</th>
<th>Patient</th>
<th>Normative</th>
<th>Z-score</th>
</tr>
</thead>
<tbody>
<tr>
<td>S-N (mm)</td>
<td>67.3</td>
<td>72.6 (3.7)</td>
<td>-2.0</td>
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<tr>
<td>S-Ba (mm)</td>
<td>36.2</td>
<td>42.4 (3.7)</td>
<td>-1.7</td>
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<tr>
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<td>51.2 (3.2)</td>
<td>-4.9</td>
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<tr>
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<td>50.4 (3.1)</td>
<td>-2.7</td>
</tr>
<tr>
<td>N-Me (mm)</td>
<td>100.6</td>
<td>112.1 (5.7)</td>
<td>-2.0</td>
</tr>
<tr>
<td>Ar-Go (mm)</td>
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<td>41.2 (3.5)</td>
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<tr>
<td>N-S-Ba (°)</td>
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<td>128.8 (4.6)</td>
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<td>N-A / FH (°)</td>
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<td>88.3 (3.1)</td>
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<td>74.3 (3.0)</td>
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<td>S-N-B (°)</td>
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<td>76.5 (3.4)</td>
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<td>7.6 (5.6)</td>
<td>-3.9</td>
</tr>
<tr>
<td>A-N-B (°)</td>
<td>-5.8</td>
<td>4.0 (2.6)</td>
<td>-3.8</td>
</tr>
</tbody>
</table>

Discussion

We described the clinical and cephalometric phenotypic features of a female patient with the syndrome of solitary maxillary central incisor and short stature, with normal growth hormone levels. The absence of the permanent maxillary lateral incisors and, more strikingly, all four mandibular incisors denotes that the developmental defect

Figure 1. Patient’s dentition on admission. Single maxillary central incisor, absent permanent lateral upper incisors, retention of primary mandibular incisors.

Figure 2. Patient’s palate, highly arched

Figure 3. Panoramic radiograph of the patient at the age of 8. Absence of the permanent maxillary lateral incisors and the four mandibular incisors

Figure 4. Lateral cephalometric radiograph of the patient. Platybasia, concave profile (see text)
of the patient, not done to any of the previously reported cases. In addition, by using the "multidimensional scaling" statistical technique, we tried to examine differences or similarities among syndromes for all characteristic cephalometric measurements found in our case, collecting data from the literature concerning such measurements in other syndromes. Unfortunately, there were not enough data for all the other syndromes in all the measurements to be included in this analysis, including five valuable cases of DeMyer (holoprosencephaly) sequence 31, which could be very informative. Finally, only six measurements: 1) anterior and 2) posterior cranial base, 3) cranial base-saddle angle, 4) maxillary length, 5) maxillary protrusion and 6) mandible protrusion, of six syndromes namely: SMCI and short stature (our case), Rieger 32, Binder 33, 34, Johanson-Blizzard 35, cerebral gigantism 36 and Down 37 were used in the analysis.

The results were clear cut. First, the model fit very well the data, that is the multidimensional scaling captured successfully the differences among syndromes in the cephalometric space; this is shown by the high coefficient of determination, R2=0.868. Second, the 2-dimensional plot (Figure 5) shows clearly that the syndrome of SMCI and short stature is quite distinct from the other five syndromes. In fact, it is more so than some other syndromes are distinct among themselves. This shows that cephalometric measurements are indeed a powerful tool in distinguishing syndromes.

Conclusion

For the future, we need more cases of the syndrome of SMCI and short stature, and cases of the A.D. form of holoprosencephaly covering the wide spectrum between the two extreme manifestations of the gene expression, i.e., holoprosencephaly and SMCI, to be cephalometrically analysed. That will, at least partly, elucidate the pathogenesis of the SMCI and short stature syndrome, whether it is simply a craniofacial dysplasia or a cerebral craniofacial dysplasia and give us information for the genetic counseling.

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