Evaluation of the severity of malocclusions in children affected by osteogenesis imperfecta with the peer assessment rating and discrepancy indexes

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Introduction: Osteogenesis imperfecta is a heritable disorder affecting bone and tooth development. Malocclusion is frequent in those affected by osteogenesis imperfecta, but this has not been studied in detail. The purpose of this study was to describe and quantify the severity of malocclusions in patients with osteogenesis imperfecta.

Methods: Articulated dental casts were obtained from 49 patients diagnosed with osteogenesis imperfecta (ages 5-19 years; 28 female) and 49 age- and sex-matched control subjects who did not have osteogenesis imperfecta. Both groups were seeking orthodontic treatment. Malocclusions were scored by using the peer assessment rating (PAR) and the discrepancy index (DI).

Results: The average United Kingdom weighted PAR scores were 31.1 (SD, 14.5) for the osteogenesis imperfecta group and 22.7 (SD, 10.7) for the control group (P <0.05). The mean United States weighted PAR scores were 32.2 (SD, 15.0) for patients with osteogenesis imperfecta and 21.6 (SD, 9.6) for the controls (P <0.05). The average modified DI scores were 29.8 (SD, 20.2) for the osteogenesis imperfecta group and 12.4 (SD, 6.8) for the control group (P <0.05). Group differences were greatest for lateral open bite (osteogenesis imperfecta group, 7.1; control group, 0.3) for the DI parameters and anterior crossbite (osteogenesis imperfecta group, 13.0; control group, 3.8 [United Kingdom]) for the PAR.

Conclusions: Both the PAR and the DI showed that malocclusions were significantly more severe in patients with osteogenesis imperfecta than in the control group. There was a higher incidence of Class III malocclusion associated with anterior and lateral open bites in patients affected by osteogenesis imperfecta. (Am J Orthod Dentofacial Orthop 2013;143:336-41)

Osteogenesis imperfecta, also known as “brittle bone disease,” is a heritable disorder of the connective tissue with a cumulative incidence of 1 in 20,000.1 Osteogenesis imperfecta is characterized by multiple fractures, muscle weakness, joint laxity, curved bones, blue sclerae, hearing loss, and dentinogenesis imperfecta. In about 90% of these patients, the disease is caused by dominant mutations in the genes that code for type 1 collagen alpha chains (COL1A1 and COL1A2).1,2 Mutations in other genes, such as CRTAP and LEPRE1, lead to severe recessive forms of osteogenesis imperfecta.2

The widely used classification of osteogenesis imperfecta by Sillence et al3 is based on clinical and radiographic criteria and mode of inheritance. It divides the disease into 4 major categories, ranging in severity from mild to lethal. Type I is the mildest form, type II is the most severe form and is lethal at birth, type III is the most severe form found in surviving patients, and type IV is of intermediate severity (between types I and III). Recently, the classification has been expanded to account for new clinical and genetic findings. Glorieux et al4,5 described osteogenesis imperfecta types V

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and VI as moderate to severe. Up to 11 types of osteogenesis imperfecta have been proposed (Table I).\(^2\)

Dental manifestations such as dentinogenesis imperfecta are well documented in osteogenesis imperfecta. In addition, several authors have studied skeletal and dental malocclusions in groups of 28 to 40 patients and reported overrepresentations of Class III malocclusions, posterior and anterior open bites, crossbites, and impacted teeth.\(^6\)-\(^14\) These reports were mainly based on clinical observations, and the severity of the malocclusions was not quantified. Also, no study has compared the severity of the malocclusions between patients with osteogenesis imperfecta and an otherwise healthy group of patients seeking orthodontic treatment.

Several specific occlusal indexes—the peer assessment rating (PAR) and the discrepancy index (DI)—are currently used to assess the severity of a malocclusion.\(^15\)-\(^21\) These indexes were developed to objectively quantify the dental malocclusions.

The PAR was developed in the United Kingdom to record the malocclusion at any stage of treatment.\(^18\) It is a rapid and accurate method of measuring dento-occlusal changes on study models.\(^22\) The validity and reliability of the PAR have been confirmed by several studies.\(^18,23,24\) Richmond et al.\(^18\) and DeGuzman et al.\(^24\) stated that a statistical weighting should be incorporated into the PAR scores to adjust them according to the population studied.

The DI was developed by the American Board of Orthodontics to study and grade cases presented for board certification.\(^15\) This index uses several criteria regarding occlusal factors, which are standardized and measured to evaluate the complexity of orthodontic cases.

### MATERIAL AND METHODS

In the past 5 years, over 100 patients with a diagnosis of osteogenesis imperfecta were referred from the Montreal Shriners Hospital to the Dental Clinic of the Montreal Children Hospital in Canada for dental treatment and assessment of their malocclusions. Forty-nine patients (ages 5-19 years; 28 female, 21 male) agreed to have orthodontic records taken for potential orthodontic treatment and matched for sex and age with the osteogenesis imperfecta group; (3) compare the PAR and the DI scores of the patients affected by osteogenesis imperfecta with those of the control group; and (4) evaluate whether the malocclusions observed in a group of patients affected by osteogenesis imperfecta were more severe than the malocclusions in a group of healthy patients seeking orthodontic treatment.

### Table I. Expanded classifications of Silence et al\(^3\) of osteogenesis imperfecta\(^1\)

<table>
<thead>
<tr>
<th>Type</th>
<th>Clinical severity</th>
<th>Typical features</th>
<th>Typically associated mutations*</th>
</tr>
</thead>
<tbody>
<tr>
<td>I</td>
<td>Mild nondeforming osteogenesis imperfecta</td>
<td>Normal height or mild short stature; blue sclerae; no dentinogenesis imperfecta</td>
<td>Premature stop codon in COL1A1</td>
</tr>
<tr>
<td>II</td>
<td>Perinatal lethal</td>
<td>Multiple rib and long-bone fractures at birth; pronounced deformities; broad long bones; low density of skull bones on radiographs; dark sclerae</td>
<td>Glycine substitutions in COL1A1 or COL1A2</td>
</tr>
<tr>
<td>III</td>
<td>Severe deforming</td>
<td>Very short; triangular face; severe scoliosis; grayish sclerae; dentinogenesis imperfecta</td>
<td>Glycine substitutions in COL1A1 or COL1A2</td>
</tr>
<tr>
<td>IV</td>
<td>Moderately deforming</td>
<td>Moderately short; mild to moderate scoliosis; grayish or white sclerae; dentinogenesis imperfecta</td>
<td>Glycine substitutions in COL1A1 or COL1A2</td>
</tr>
<tr>
<td>V</td>
<td>Moderately deforming</td>
<td>Mild to moderate short stature; dislocation of radial head; mineralized interosseous membrane; hyperplastic callus; white sclerae; no dentinogenesis imperfecta</td>
<td>Unknown</td>
</tr>
<tr>
<td>VI</td>
<td>Moderately to severely deforming</td>
<td>Moderately short; scoliosis; accumulation of osteoid in bone tissue; fish-scale pattern of bone lamellation; white sclerae; no dentinogenesis imperfecta</td>
<td>Homozygous SERPINF1 mutations</td>
</tr>
<tr>
<td>VII</td>
<td>Moderately deforming</td>
<td>Mild short stature; short humeri and femora; white sclerae; no dentinogenesis imperfecta</td>
<td>Homozygous CRTAP mutations</td>
</tr>
</tbody>
</table>

*May or may not be detectable in a given patient.
Dental evaluations, extraoral and intraoral photographs, and alginate impressions for study models were taken. Intraoral radiographs, panoramic views, and cephalometric images were usually taken when possible. The plaster models were trimmed according to the American Board of Orthodontics standards and placed into occlusion. Bite registrations were obtained by using both Aquasil Easy Mix Putty (Dentsply International, York, Pa) and Base Plate Modeling Wax 101 (Carmel, Montreal, Quebec, Canada).

The control group consisted of patients seeking orthodontic treatment at 2 private orthodontic offices and who were matched with the patients in the osteogenesis imperfecta group for age, sex, and ethnicity. The first matching control from a random selection was paired with each patient with osteogenesis imperfecta. We preferred a control group of patients seeking orthodontic treatment because the 49 patients affected by osteogenesis imperfecta were seeking orthodontic treatment as well.

All study models were examined and measured by 2 dentists (J.R. and S.S.) who followed the respective protocols of the PAR and the DI. The scores were entered separately, and the results compared every 10 subjects. If the 2 clinicians recorded entries that differed by more than 1 point, the measurements were taken again, and the subject was discussed until both clinicians accepted the same results. A high interrater agreement was found (intraclass correlation coefficient, 0.996; 95% confidence interval, 0.986-0.999).

The PAR index uses 7 criteria: tooth alignment (referring to dental crowding), right and left buccal segment relationship (sagittal, vertical, and transverse assessments), overjet, overbite, and centerline (midline discrepancies). Each difference from the norm has points attributed to the severity of the discrepancy. Once tabulated and weighted according to the United States (US) or United Kingdom (UK) weightings, an overall score for the malocclusion is calculated.

The DI as related to dental casts includes 7 parameters: overjet, overbite, open bite, crowding, occlusal relationship, crossbite, and a category called “other” that includes such anomalies as missing teeth. Similarly to the PAR, scores are assigned and tabulated to reflect the malocclusion.

Angle molar relationship, according the Angle classification, was also recorded. A flexible plastic ruler, thinned for the first 25 mm, and a caliper (Hu-Friedy, Chicago, Ill) were used for measurements.

**Statistical analysis**

Differences in mean scores for the DI between the osteogenesis imperfecta and the control groups were compared as follows. For the US and UK weighted PAR index, the following categories were combined by adding the following scores: (1) upper right, upper anterior, and upper left segments into the “upper segment” category; (2) lower right, lower anterior, and lower left segments into the “lower segment” category; (3) right and left buccal occlusion anteroposterior; (4) right and left buccal occlusion vertical; and (5) right and left buccal occlusion transverse.

The differences between groups for each category were compared by using independent-sample t tests. All analyses were performed with statistical software (PASW version 18; SPSS Inc, Chicago, Ill). A P value less than 0.05 was considered significant. Intraclass correlation coefficients were calculated to test the interrater agreement between the 2 raters, based on the scores for a set of 10 patients.

**RESULTS**

Examples of malocclusions in patients with osteogenesis imperfecta are shown in Figures 1 and 2. The osteogenesis imperfecta and the control groups were identical for sex distribution and were similar with regard to age (mean, 10.7 years; SD, 3.3 years in the osteogenesis imperfecta group; mean, 10.6 years; SD, 3.0
The Angle molar classification differed markedly between the 2 groups. Among the patients affected by osteogenesis imperfecta, 12 (25%) had Class I, 9 (18%) had Class II, and 28 (57%) had Class III malocclusions. In the control group, 16 (33%) had Class I, 31 (63%) had Class II, and 2 (4%) had Class III malocclusions (*P < 0.001 by the chi-square test for the difference in the distribution between the groups).

The PAR scores in the osteogenesis imperfecta group ranged from 2 to 64 (UK weighting) and from 3 to 69 (US weighting); the corresponding results in the controls ranged from 5 to 45 (UK weighting) and from 1 to 49 (US weighting). For both the UK and the US weighted PAR scores, the results were significantly higher in patients affected by osteogenesis imperfecta (Table II).

The osteogenesis imperfecta group had a higher score for buccal occlusion in the vertical dimension (posterior open bite), buccal occlusion, transverse (posterior crossbite), anterior crossbite, and anterior open bite, whereas the control group had higher scores for overjet and overbite. No significant group differences were found for the “displacement scores” (corresponding to dental crowding), the “anteroposterior buccal occlusion” (where the interdigitation of the maxillary and mandibular lateral segments is assessed), and the “centerline” (where the midline discrepancy in relation to the mandibular incisors is measured).

The DI scores ranged from 3 to 83 in the osteogenesis imperfecta cohort and from 3 to 34 in control group (Table III). Analysis of the individual DI components showed that the osteogenesis imperfecta group had higher scores for anterior and posterior lingual crossbites, anterior and posterior open bites, Angle occlusion classes, and “others” whereas the control group had higher scores for overjet and overbite. Differences in crowding and buccal posterior crossbites were not statistically significant.

When the total scores were tabulated for the PAR (US) and the DI, 53% and 39% of the patients affected by osteogenesis imperfecta scored over 31 points, whereas 10% and 2% of the control group reached that level (*P < 0.001 by the chi-square test for both parameters).

**DISCUSSION**

In this study, we observed that osteogenesis imperfecta patients have more severe malocclusions than did the control group of otherwise healthy patients seeking orthodontic treatment. Lateral open bites, a rare finding in the general population, were particularly prevalent in the osteogenesis imperfecta patients.

### Table II. Results of the UK and US weighted PAR scores

<table>
<thead>
<tr>
<th>PAR parameters</th>
<th>OI UK</th>
<th>Control UK</th>
<th>OI US</th>
<th>Control US</th>
</tr>
</thead>
<tbody>
<tr>
<td>Total</td>
<td>31.1 (14.5)*</td>
<td>22.7 (10.7)*</td>
<td>32.2 (15.0)*</td>
<td>21.6 (9.6)*</td>
</tr>
<tr>
<td>Upper segment</td>
<td>5.5 (5.4)</td>
<td>3.6 (3.4)</td>
<td>5.5 (5.4)</td>
<td>3.6 (3.4)</td>
</tr>
<tr>
<td>Lower segment</td>
<td>2.7 (3.4)</td>
<td>2.8 (3.0)</td>
<td>0.0 (0.0)</td>
<td>0.0 (0.0)</td>
</tr>
<tr>
<td>Buccal occlusion: anteroposterior</td>
<td>1.3 (1.5)</td>
<td>1.4 (1.4)</td>
<td>2.5 (2.9)</td>
<td>2.8 (2.7)</td>
</tr>
<tr>
<td>Buccal occlusion: vertical</td>
<td>0.7 (0.9)*</td>
<td>0.0 (0.1)*</td>
<td>1.5 (1.9)*</td>
<td>0.0 (0.3)*</td>
</tr>
<tr>
<td>Buccal occlusion: transverse</td>
<td>3.7 (2.2)*</td>
<td>1.1 (1.8)*</td>
<td>7.4 (4.4)*</td>
<td>2.3 (1.6)*</td>
</tr>
<tr>
<td>Overjet (mm)</td>
<td>0.1 (0.9)*</td>
<td>6.0 (7.2)*</td>
<td>0.1 (0.7)*</td>
<td>5.0 (6.0)*</td>
</tr>
<tr>
<td>Anterior crossbite</td>
<td>13.0 (9.6)*</td>
<td>3.8 (7.0)*</td>
<td>10.8 (8.0)*</td>
<td>3.2 (5.8)*</td>
</tr>
<tr>
<td>Overbite (mm)</td>
<td>0.2 (0.6)*</td>
<td>1.8 (1.9)*</td>
<td>0.3 (0.9)*</td>
<td>2.7 (2.8)*</td>
</tr>
<tr>
<td>Open bite</td>
<td>1.6 (2.1)*</td>
<td>0.4 (1.4)*</td>
<td>2.4 (3.2)*</td>
<td>0.7 (2.1)*</td>
</tr>
<tr>
<td>Centerline</td>
<td>2.2 (3.3)</td>
<td>1.7 (2.6)</td>
<td>1.7 (2.5)</td>
<td>1.3 (1.9)</td>
</tr>
</tbody>
</table>

Results are given as means (SD).

*Significant group differences, *P < 0.05, with independent t tests.

### Table III. Results of the DI scores

<table>
<thead>
<tr>
<th>DI parameters</th>
<th>OI</th>
<th>Control</th>
</tr>
</thead>
<tbody>
<tr>
<td>Total</td>
<td>29.8* (20.2)</td>
<td>12.4* (6.8)</td>
</tr>
<tr>
<td>Anterior crossbite</td>
<td>5.6* (7.0)</td>
<td>0.7* (1.4)</td>
</tr>
<tr>
<td>Overjet</td>
<td>0.1* (0.2)</td>
<td>1.6* (1.7)</td>
</tr>
<tr>
<td>Overbite</td>
<td>0.2* (0.8)</td>
<td>1.3* (1.6)</td>
</tr>
<tr>
<td>Anterior open bite</td>
<td>3.7* (6.0)</td>
<td>0.8* (3.4)</td>
</tr>
<tr>
<td>Lateral open bite</td>
<td>7.1* (11.8)</td>
<td>0.3* (0.9)</td>
</tr>
<tr>
<td>Crowding</td>
<td>3.6 (2.9)</td>
<td>3.1 (2.5)</td>
</tr>
<tr>
<td>Occlusion</td>
<td>5.3* (5)</td>
<td>2.9* (2.9)</td>
</tr>
<tr>
<td>Lingual posterior crossbite</td>
<td>2.0* (1.7)</td>
<td>0.5* (1.1)</td>
</tr>
<tr>
<td>Buccal posterior crossbite</td>
<td>0 (0)</td>
<td>0.4 (1.7)</td>
</tr>
<tr>
<td>Other</td>
<td>2.0* (2.7)</td>
<td>0.5* (1.3)</td>
</tr>
</tbody>
</table>

Results are given as mean (SD).

*Significant group differences, *P < 0.05, with independent t tests.
Most of our population affected by osteogenesis imperfecta had types III, IV, and VI, which are the most severe (84% of our osteogenesis imperfecta group). These patients are much more closely followed and have challenging needs that are usually not compatible with care in conventional dental clinics. In the general population, osteogenesis imperfecta type I, the milder form of the syndrome, is the most prevalent but represented only 16% of our sample.1 This low number is explained by the fact that type I patients are usually cared for in conventional dental offices and not in specialized centers. The dentition is more harmonious, and dentinogenesis imperfecta is not as prevalent; consequently, families often are not aware of dental problems and do not seek specialized dental help.

The mean PAR and DI scores for the control group in this study were similar to those in previous studies.25-27 Less than 10% of the control group reached a high PAR or DI score (>31), whereas over 40% of the group affected by osteogenesis imperfecta did. These numbers emphasize the severity of the malocclusions in these patients.

Both indexes used in this study agreed about the statistical differences of each parameter studied, except for the sagittal relationship of the posterior teeth. This difference was caused by the respective methods of scoring discrepancies. As long as the interdigititation is deemed acceptable, the PAR does not acknowledge the difference between Class I, Class II, and Class III, whereas the DI does. The group affected by osteogenesis imperfecta was mainly represented by Class III (57%) molar relationships, whereas the control group was mainly represented by Class II (63%) molar relationships. Therefore, even if the malocclusions had totally different classification, the PAR did not reflect accurately the sagittal discrepancies between the group affected by osteogenesis imperfecta and the control group.

The PAR system includes a score for dental alignment needs. It is called "displacement scores" and gives a value to the distance between 2 contact points of adjacent teeth.16 Even though the DI uses a different measuring technique, the results of both indexes for dental crowding showed no significant difference between the 2 groups. Dental crowding is usually the most frequent kind of malocclusion; in this respect, the osteogenesis imperfecta patients are no different from any group in need of orthodontic treatment.

Midline discrepancies were measured by the PAR but excluded by the DI. The PAR did not indicate a statistically significant difference between the 2 groups. Since the osteogenesis imperfecta syndrome does not affect facial symmetry, it was expected to obtain similar midline discrepancy scores.

Both indexes showed a high incidence of lateral open bites in the osteogenesis imperfecta group. The PAR scores the absence (score of 0) or the presence (score of 1) of an open bite, but the DI takes into consideration the number of teeth and the amount of opening in millimeters. In this particular feature, the data obtained by the PAR remain qualitative and not informative regarding the severity of the observation, whereas the data from the DI quantify the severity of the problem (7.1 in the osteogenesis imperfecta group; 0.3 in the control group). Anterior crossbites and open bites were more frequent in the patients affected by osteogenesis imperfecta in both indexes: the PAR measured their severity by counting the number of teeth involved in crossbite and measured only the maximum distance between the 2 incisors. Again, the DI gives a better quantitative measurement by calculating the millimeters between each tooth in crossbite as well as in open bite.

Both indexes recognized the severity of posterior crossbites in the group affected by osteogenesis imperfecta. This finding is to be expected because, even in a normal population, there is a high correlation between Class III malocclusion and posterior crossbite. The PAR registered a higher average score because it incorporates what is called "crossbite tendency," whereas the DI considered only "true" crossbites.

Overbite and overjet were the only parameters where the control group had a significantly higher level of severity than the group affected by osteogenesis imperfecta. This finding can be attributed to the much larger representation of Class II subjects in our control group. A Class II malocclusion is usually associated with greater overbites and overjets. The measurements were quite similar in both indexes, but the weightings in the PAR attached a higher importance to overjet, resulting in an increased mean score. On the other hand, it has been shown that an overwhelming majority of patients affected by osteogenesis imperfecta with malocclusions have Class III maloclusions. Class III malocclusions are rarely associated with large increases in overjet and overbite.

Based on our results, a relative mandibular dentoalveolar prognathism is present in most of the patients severely affected by osteogenesis imperfecta. Previous reports demonstrated that a skeletal Class III pattern can be due to a lack of maxillary development, a mandibular prognathism, or a combination of both.5,7,12 A recent cephalometric study demonstrated that vertical underdevelopment of the dentoalveolar structures and the condylar process was the main reason for the relative mandibular prognathism in osteogenesis imperfecta.13 The authors also noted a primary growth defect of the cranial base and condylar cartilages.
similar to the ones found in endochondrally forming long bones in severe types of osteogenesis imperfecta. It is reasonable to speculate that the relative prognathism observed in patients affected by osteogenesis imperfecta is due to a lack of anteroinferior displacement of the maxilla caused by a primary growth defect at the cranial base combined with vertical underdevelopment of the dentoalveolar structures and the condylar process. This results in an anticlockwise rotation of the mandible, hence the Class III malocclusion.

**CONCLUSIONS**

The results of this study showed that patients more severely affected by osteogenesis imperfecta share common characteristics: severe malocclusions, Class III malocclusions, and anterior and posterior open bites and crossbites.

Both the PAR and the DI showed that the malocclusions in the osteogenesis imperfecta group were significantly more severe than those of the control group. Future studies will focus on better understanding of the etiology and the development of these malocclusions.

We thank Dr. Claudia Giambattistini for supplying control study models.

**REFERENCES**