INTERNATIONAL COUNCIL of OPHTHALMOLOGY
WOC 2020
WORLD OPHTHALMOLOGY CONGRESS
VIRTUAL 26 – 29 JUNE

CORNEAL TOMOGRAPHY IN OSTEOGENESIS IMPERFECTA

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INTRODUCTION

Osteogenesis Imperfecta (OI) is a rare genetic disorder with musculoskeletal manifestations and some ocular features, such as the presence of **blue sclerae, corneal thinning, altered corneal biomechanics and rare reports of keratoconus (KC)[1,2]**. Corneal tomography is essential for KC screening and a BAD_D score over 1,6 and 2,6 is highly sensitive and specific for the detection of subclinical KC and keratoconic corneas, respectively[3-6]. **This study aims to explore the corneal tomographic characteristics of OI patients.**

METHODS

- Cross sectional study of 31 adult OI patients recruited form the Portuguese OI Association;
- All subjects underwent ophthalmological evaluation and corneal tomography (Pentacam HR, Oculus, Germany);
- Further clinical data was retrieved from patient files;
- Statistical analysis performed with Graphpad Prism 8 (San Diego, CA, USA) and p-value of < 0.05 for statistical significance was used.

RESULTS

Our sample consisted of **31 OI patients**, two of which were subsequently excluded due to poor exam cooperation. The **remaining 29 patients** (22 female), were adult caucasians with an average age of 42±15 years. **Most patients (n=18; 62%) had OI type I disease**, while the remaining patients had less frequent phenotypes, such as OI type III (n=3), type IV (n=3), type V (n=1), type VI (n=2) and type VII (n=1), and one patient still unclassified. Our main results showed:

- **Average minimal pachymetry was 478±54µm, which is significantly lower than the reference range of 550±30µm[5,8] (p<0,001);**
- Most tomographic indices used for KC diagnosis are significantly deviated in OI patients compared with the normal range, particularly in OI type I (table);
- **Twenty-two patients (76%) had at least one eye with a BAD > 1,6 and nine patients (31%) had at least one eye with a BAD > 2,6;**
- Only two patients (7%) of these had clinical signs of KC such as Munson's sign, Fleischer ring or Vogt's striae.

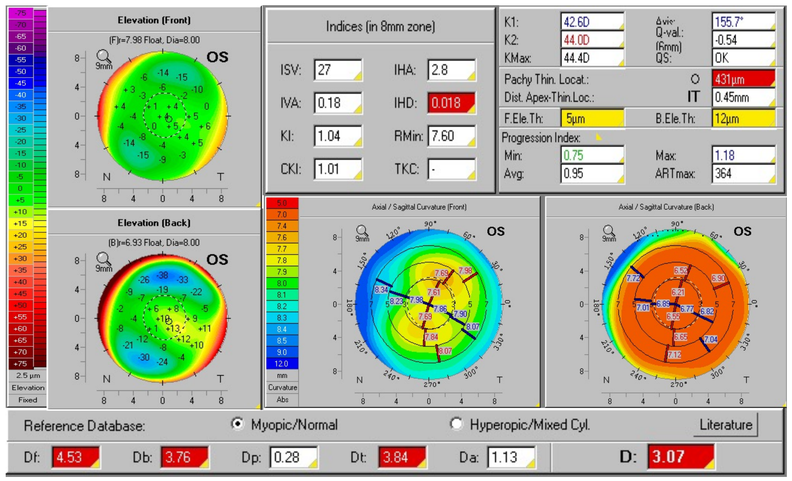

Figure. **Cropped portions of Pentacam displays from the left eye of patient 2**, indluding front and back elevation maps, curvature maps and KC indices. This was a 37 year-old male with no biomicroscopic signs of corneal ectatic disease. However, corneal tomography shows a markedly thin cornea, slightly abnormal front and back elevations, normal keratometry and a BAD_D score of 3.07, indicating KC.

KC indices	OI (mean±SD)	OI type I (mean±SD)	Normal range[3-8] (mean±SD)	OI vs. Normal p-value
IVA	0.22±0.15	0.25±0.18	0.10±0.0 - 0.21±0.08	0,5998
ISV	**26.9±13.9**	**28.8±15.9**	**19.0±7.00 - 22.5±7.66**	**0,0204***
IHA	**8.01±6.36**	**8.80±5.57**	**3.39±2.80 - 5.33±4.59**	**0,0022***
IHD	**0.020±0.015**	**0.021±0.016**	**0.006±0.003 - 0.01±0.03**	**<0,0001***
BFS Front Thin 8mm (µm)	3.14±3.70	4.06±4.30	1.57±2.04 – 3.0±2.0	0,7773
BFS Back Thin 8mm (µm)	**12.0±8.57**	**14.0±10.1**	**3.55±4.18 - 4.1±4.1**	**<0,0001***
Minimal pachymetry (µm)	**478±53**	**464±40**	**537±31 - 549±36**	**<0,0001***
PPIavg	1.016±0.202	1.010±0.163	0.85±0.11 -0.99±0.14	0,3275
ARTmax	**389±100**	**378±83.5**	**445±87 - 527±91**	**<0,0001***
K avg	43.8±1.98	43.5±1.99	43.8±1.4	0,9056
K max	45.4±2.30	45.0±1.97	44.6±1.5 - 45.4±1.5	0,9818
BAD_D	**2.22±1.53**	**2.55±1.67**	**0.43±0.57 - 1.03±0.70**	**<0,0001***

Table. **KC indices in OI patients vs. Normal range**. ARTmax, maximum Ambrósio relational thickness; BAD, Belin/Ambrosio Enhanced Display; BFS, best-fit sphere; IHA, index of height asymmetry; IHD, index of height decentration; ISV, index of surface variance; IVA, index of vertical asymmetry; K max and avg, maximum and average keratometry; PPIavg, Average pachymetry progression. *Indicates statistically significant difference.

CONCLUSIONS

Our study show that OI patients often display corneal tomographic abnormalities consistent with ectatic disease. Corneal thickness is significantly reduced and over three-quarters of our patients had at least one eye with a BAD score indicative of subclinical KC and approximately a third had at least one eye with a BAD indicative of overt KC. However, few of our patients showed clear clinical signs of KC disease. Some KC indices, such as the ARTmax and BAD, use pachymetric information and thus could be skewed by the low pachymetric readings in OI patients.

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