Osteogenesis Imperfecta (OI), is a genetically heterogeneous connective tissue disorder associated with skeletal fragility, deformity, and growth deficiency.

Intravenous bisphosphonate therapy is the mainstay of medical treatment of this condition.

Majority of our patients (55.2%) were OI type III, followed by types I and IV (17.2% each). 73% of our patients had a mutation in the collagen gene.

Response to pamidronate therapy

- The fracture rate decreased at all available time points for patients on pamidronate, compared to the year prior to treatment. (Chart 1)
- There was no significance difference of height pre and post pamidronate therapy. There was no deterioration of height SDS with time among these group of patients. (Chart 2)

Pharmacogenetic effects of bisphosphonate treatment

- Patients who had quantitative mutations had a milder phenotype as compared to those with qualitative mutations.
- There was no significant difference when comparing individuals who are COL1A1 positive with those who are COL1A2 positive as regards fracture rate and height SD.

CONCLUSIONS

- Patients with haploinsufficiency mutations had a milder phenotype as compared to those with qualitative mutations.
- In the group of patients with helical mutations, the type of alpha chain affected did not influence the fracture rate.
- Cyclic pamidronate administration reduced the fracture rate effectively in patients with OI.

REFERENCES