Living with Osteogenesis Imperfecta and Reaching the Golden Years

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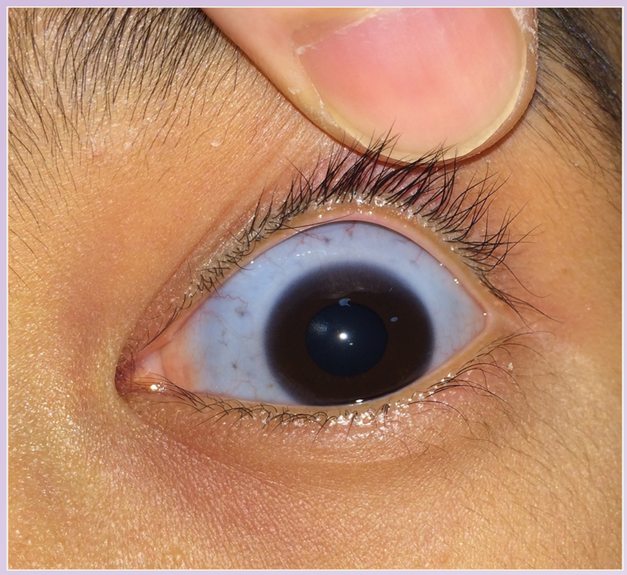
Background

- Osteogenesis Imperfecta (OI) or "brittle bone disease," is a genetic connective tissue disorder with varied disease presentation. It is associated with multiple lifetime fractures secondary to minimal trauma.
- Several types have been identified (Table). The severity ranges from benign early-onset osteoporosis and joint laxity in Type I to lethality in the perinatal period in Type 2.
- Current treatments include minimizing fractures and mitigating associated complications.
- Lifespan is dependent on clinical features and OI type.
- Patients who survive into adulthood often experience limited functionality, particularly with more severe disease burden.
- We present a fascinating case of a patient with OI and relatively high quality of life despite marked phenotypic severity who is approaching retirement.

Example of blue sclera, typical of specific OI types (1)



Case

This is a 65-year-old lady with reportedly type III osteogenesis imperfecta, type II diabetes mellitus and asthma. Her mother and three of nine siblings are affected with OI as well. She was diagnosed at six years of age after sustaining a fracture upon falling out of bed. Some of her phenotypic features include blue sclera (similar to what is depicted in the figure), short stature (4'11" currently), early tooth loss and hearing loss.

The patient's mother placed her in a handicapped school, anticipating the physical impairments that would accompany her disease. At school, she was taught strategies on minimizing injuries including "how to fall" in order to avoid fracture. Despite these efforts, she sustained 12 lifetime fractures, with the majority occurring between the ages of six and 18. She has never required any surgical intervention but has required casts.

Currently, she follows with a primary care physician as well as an endocrinologist. She has required temporary bisphosphonate therapy following a tailbone fracture at the age of 60. She continues to lead a highly functional lifestyle with minimal maintenance therapy. She is self-confident, lives alone and has a steady job.

OI types and features (2) . AR- autosomal recessive , AD- autosomal dominant

Type	I[a]	II-A[b]	II-B[b]	III[a]	IV[a]	V[c]	VI[d]
Inheritance	AD	AD	AD/AR[e]	AD/AR[f]	AD/AR[g]	AD	AR
Severity	mild	perinatal lethal	severe	moderate-mild	moderate	moderate	moderate
Congenital fractures	no	yes	yes	usually	rarely	no	no
Bone deformity	rarely	very severe	severe	moderate-severe	mild-moderate	moderate	moderate
Sclerae	predominantly blue[h]	dark blue[i]	dark blue[i]	blue/grey/white[j]	normal-grey[k]	normal	normal
Stature	normal[l]	severely short[m]	severely short[n]	very short[o]	variable short[p]	variable	mild short stature
Joint hypermobility	yes[q]	yes	yes	yes[q]	variable	variable	variable[r]
Hearing loss	present in ~60% of cases[s]	NA	NA	common[t]	present in ~42% of cases[u]	no	no
Dentinogenesis imperfecta (DI)	variable[v]	NA	NA	yes[w]	variable	no	no
Respiratory complications	no	yes[x]	yes[x]	yes[y]	no	no	no
Neurological complications	no	NA	NA	yes[z]	no	no	no

✓ Resources are available for patients to refer to on the OI foundation website. Guidance on daily care is provided to patients.

Discussion

- OI can be extremely disabling in patients with severe presentations. Patients with severe phenotypic features have limited functionality due to higher likelihood of recurrent fractures.
- Our patient is an unusual case of severe OI, with relatively few complications and long term sequelae of her multiple fractures. She was able to maintain good quality of life, is confident and approaching retirement. This is likely due to being taught precautionary skills at an early age, a healthy lifestyle and success in minimizing the morbidity that is associated with OI.
- Some individuals with Type III OI have severe, often fatal, respiratory problems due to rib cage and spine deformities. Others may lead a near-average life span.
- Patients with non-lethal variants should get genetic counseling. Affected children will benefit from early lifestyle modifications and special learning skills to avoid of trauma so they can enter adulthood and lead highly functional lives.
- Psychological evaluation or treatment may be warranted.
- When caring for patients with OI, a multidisciplinary approach is needed (surgeons, physicians, PT/OT, behavioral health and dietitians).

References:

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3- Osteogenesis Imperfecta Foundation https://oif.org/information-center/about-oi/

4- Marr Caroline et al. Managing the patient with osteogenesis imperfecta: a multidisciplinary approach. J Multisicip Healthc 2017;10:145