

Hearing Loss

Deaf-guy

Hearing loss is progressive in these patients and can begin at an early age. Up to 50% of patients are affected by this. The pathophysiology is similar to otosclerosis and is an audio conduction pathology.

Dental Imperfections

Bad teeth

Commonly, patients develop dental imperfections with this disease, due to a lack of dentin. Findings include obliterated pulp cavities, opalescent teeth, and constricted coronal-radicular junctions.

Blue Sclera

Blue-eyed Skull

As type I collagen formation is defective, the sclera of the eye is much thinner than normal. Because of this, the underlying choroidal veins show through, giving the eye a blue or sometimes grey color.