

**Table 1**

**Osteogenesis imperfecta: classification and characteristics of the four most common types**

| Classification | Clinical characteristics                                                                                                                             | Type 1 collagen abnormality                                                                          |
|----------------|------------------------------------------------------------------------------------------------------------------------------------------------------|------------------------------------------------------------------------------------------------------|
| Type I         | Fractures, osteopaenia, blue sclera, deafness, dentinogenesis imperfecta in some children                                                            | Autosomal dominant (AD). Reduced amounts of structurally normal collagen                             |
| Type II        | Lethal; fractures in utero, soft cranium, short limbs, dark sclera                                                                                   | AD or autosomal recessive (AR). Structural mutation in one of the collagen chains. Abnormal collagen |
| Type III       | Severe. Progressively deforming, fractures at birth. Very short stature, severe scoliosis. Triangular facies, grey sclera, dentinogenesis imperfecta | AD<br>As type II                                                                                     |
| Type IV        | Moderately severe. Similar features to III but less severe. White or grey sclera                                                                     | AD<br>As type II                                                                                     |