

# **Student Essay on Osteogenesis Imperfecta**

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# Essay

"Osteogenesis Imperfecta..."

Osteogenesis imperfecta (OI) is a genetic disorder characterized by bones that break easily, often from little or no apparent cause. There are at least four recognized forms of the disorder, representing extreme variation in severity from one individual to another. For example, a person may have just a few or as many as several hundred fractures in a lifetime. While the number of people affected with OI in the United States is unknown, the best estimate suggests a minimum of 20,000 and possibly as many as 50,000.

## Diagnosis

OI is caused by a genetic defect that affects the body's production of collagen. Collagen is the major protein of the body's connective tissue and can be likened to the framework around which a building is constructed. In OI, a person has either less collagen than normal, or a poorer quality of collagen than normal--leading to weak bones that fracture easily.

It is often, though not always, possible to diagnose OI based solely on clinical features. Clinical geneticists can also perform biochemical (collagen) or molecular (DNA) tests that can help confirm a diagnosis of OI in some situations. These tests generally require several weeks before results are known, and approximately 10 to 15 percent of individuals with mild OI who have collagen testing, and approximately 5 percent of those who have genetic testing, test negative for OI despite having the disorder.