**Osteogenesis Imperfecta (OI)**

Osteogenesis Imperfecta (OI) is a rare genetic disorder which affects 1 in 10,000 people. It is characterized by fragile bones which lead to a lifetime of severe fractures. Currently there is no cure for OI as no therapy can target the primary OI problem, which is the reduced or abnormal collagen produced by bone cells.

The OI research group in VUmc, led by dr. Dimitra Micha and dr. Gerard Pals, has identified several genes responsible for OI. Their aim is to identify more genes in order to complete the genetic panel for OI.

In 2013 we discovered that the gene *PLS3* causes fractures and fragile bones. We currently investigate the mechanism by which T-plastin, which is produced by the gene *PLS3*, affects the function of bone cells. Research on bone cells is hindered by the invasiveness of bone biopsies. VUmc has developed a novel method of osteogenic transdifferentiation to make bone cells (osteoblasts) from the easily acquired skin cells of OI patients. With this technique we can research the molecular pathology of OI. These osteoblasts also serve as a screening platform to find new therapies.

The generous contribution of the Horstingstuit foundation enables us to perform unique research to find an effective treatment for OI. A number of substances which lead to increase of collagen synthesis will be examined *in vitro* (osteoblast culture) in cells of OI patients with various mutations. Our researchers will also work on repair of the mutated cells of OI type III/IV patients. Further, the effectiveness of the potential medicines will be examined *in vivo* (mouse models). The results of this research can contribute to an effective therapy for OI and improvement of quality of life for OI patients. *This research could not be realized without the kind support of the horstingstuit-foundation.*