MSc graduation project: Bone microarchitecture and strength in osteogenesis imperfecta

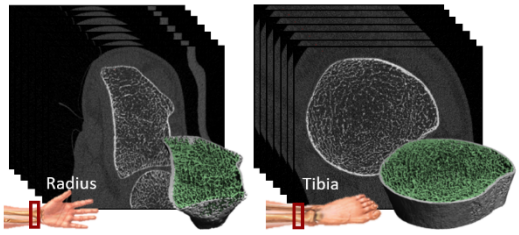
Osteogenesis imperfecta (OI) is a rare and highly heterogeneous condition, of which one characteristic is the increased fracture risk due to bone structural and material defects. Detailed data on bone structure in OI are scarce. High-Resolution peripheral Quantitative CT (HR-pQCT) enables *in-vivo* and non-invasive assessment of bone microarchitecture and strength, and we have obtained a large HR-pQCT dataset of 118 adults with OI. Standard analyses have already been performed and comparisons with normative data been made to study bone microarchitecture and strength in this cohort. Several questions still need to be answered and other questions have arisen, including:

- We observed large empty spaces in the inner trabecular compartment in part of the study participants, and we would like to know whether and to what extent the prevalence and size of these void volumes is similar as or different from void volumes in non-OI populations. An void detection algorithm has recently been published and applied to a non-OI group, and it may be applied to our cohort to enable comparison of the void characteristics in the published non-OI cohort and our OI-cohort. We also developed an algorithm for void detection, in which different choices were made regarding e.g. void definition as compared to the published algorithm. The difference in detection performance between the algorithms is yet unknown and may be studied.
- OI is divided into subgroups, of which type I is the largest. Yet, there is large genetic and clinical heterogeneity within subgroups. We would like to know whether there may be distinguishable HR-pQCT phenotypes within OI type I and if so, whether these phenotypes are associated with clinical and genetic characteristics. An approach to study this could be clustering.
- For our dataset, we have obtained HR-pQCT scans using the 'standard' scan protocol that does not take into account extremity length and an extremity-length-dependent scan protocol for each of the study participants. The latter was additionally used as shorter stature can be present in OI, which would influence scan location and thereby limit normative comparison when using the standard scan protocol. However, given the subjectivity of measuring extremity length, study is needed to determine whether use of the extremity-length-dependent scan protocol in OI leads to differences in HR-pQCT parameters from the standard scan protocol.

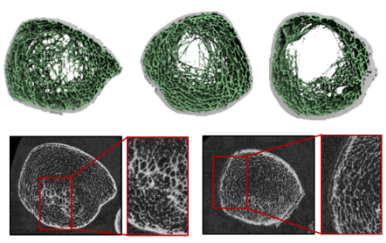
The MSc graduation project can include one or more of these projects.

Interest or more information? Contact Melissa Bevers (m.s.a.m.bevers@tue.nl)!

HR-pQCT scanner



Examples of HR-pQCT scans



Inhomogeneities observed in OI