

The SKELETOME Project: Towards a community-driven knowledge curation platform for Skeletal Dysplasias

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The Nosology of Genetic Skeletal Disorders represents the main hub for structuring and retrieving key information about skeletal dysplasias. It provides a classification of the disorders, based upon a shared understanding among the experts, in addition to a shallow grouping by common clinical-radiographic characteristics and/or molecular disease mechanism.

In parallel, several systems enabling case studies management and fostering knowledge exchange between domain experts have been developed (e.g., ESDN). Their main role is to promote diagnosis support via interactive discussions. However, their functionalities rise only to the level of a discussion forum and lack any real support for advanced features, like, retrieving information about similar cases. In addition, while the knowledge is intrinsically present in the communication process, novel findings cannot be directly reflected in the Nosology, due to its inflexible nature.

The continuous adoption of Semantic Web technologies now allows us to transform the Nosology into a more dynamic and well-grounded formalism. In this new context, the SKELETOME project aims at developing an ontology for the bone dysplasias domain, to enable knowledge consolidation and to create a comprehensive overview of the domain by adjoining several aspects describing its inherent complexity.

The SKELETOME project also plans to build an ontology-based community-driven knowledge curation platform that will enable collaborative input, sharing and re-use of data and information among experts. The goal is to provide a central access point to a rich skeletal dysplasia knowledge base, supported by low-level features, such as user and group-based access and privacy control. At the same time, from a high-level perspective, the anonymised pool of case studies will enable statistical inference for knowledge discovery purposes or computer-assisted diagnosis. Finally, the use of the ontology as foundational building block will lead a more straightforward and quicker incorporation of novel discoveries into the overall bone dysplasia domain knowledge.