Bone2Gene

Artificial Intelligence for Diagnosing and Monitoring Rare Bone Diseases

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Bone2Gene is a start-up project at University Hospitals Bonn and Magdeburg

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http://www.bone2gene.org

Overview

According to the most recent Nosology of Genetic Skeletal Disorders [1], there are over 700 known rare bone diseases (RBDs) that involve over 500 different genes. Though these diseases are individually rare, they collectively affect a large number of individuals. As with most rare diseases, diagnosing RBDs is inherently challenging, often requiring extensive time and multiple clinical visits, a process that can be frustrating for patients. Effective treatment hinges on precisely identifying the type of RBD, emphasizing the importance of addressing the current diagnostic gap. Moreover, accurate diagnosis can have economic and societal impact, as available therapies are often highly specific and may involve highly variable access and costs [2].

Goal

Bone2Gene[1] aims to revolutionize access to effective treatment methods and change the diagnostics journey using innovative artificial intelligence. Experts in the fields of big data, Artificial Intelligence (AI), medical genetics, pediatric endocrinology and radiology from the universities of Bonn and Magdeburg as well as collaborators from different centers around the world will work together to build a comprehensive database and train the Bone2Gene AI to recognize and distinguish patterns in medical imaging. In the so-called next-generation phenotyping, modern computational techniques, particularly AI-based methods, are employed for discovering new features and training algorithms that can recognize the patterns associated with different genetic diseases. Studies such as GestaltMatcher [3] and Eye2Gene [4] have demonstrated the success of AI in learning the characteristic facial and retinal features of a large number of rare genetic diseases and in accelerating the diagnostic process.

Initially, Bone2Gene AI will concentrate on training with dorsopalmar radiographs of hands and wrists from various RBDs. This method was chosen as hand X-rays for bone age assessment are routinely performed on children suspected of bone anomalies. In later phases, Bone2Gene will expand its training to encompass radiographs from additional body parts.

[1] Funded by the German Federal Ministry of Education and Research:
https://www.go-bio.de/gobio/de/gefoerderte-projekte/gobio-initial/_documents/bone2gene.html

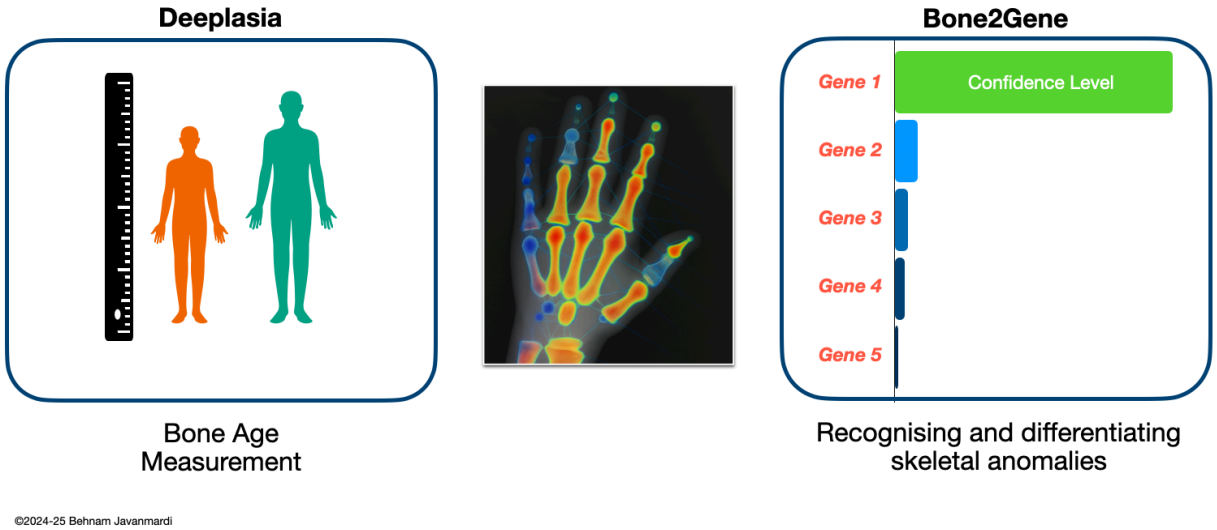

Figure 1: AI for skeletal dysplasias.

Technical Summary

As groundwork for Bone2Gene, we have already developed a bone age assessment AI (Figure 1, left) called Deeplasia[2] and have specifically validated it on a cohort of seven different skeletal dysplasia [5]. For developing Bone2Gene (Figure 1, right), we start with our Deeplasia's model weights and we leverage state-of-the-art deep learning (DL) architectures and advanced computer vision techniques to develop a multi-class classifier of different RBDs. Our current data collection includes around 3000 X-rays from over 20 different RBDs. We are expanding this data set via our global network.

The data are collected in the form of pseudonymised hand radiographs together with a set of meta information, mainly patient's sex (male/female), the confirmed diagnosis (e.g. achondroplasia with a mutation in the FGFR3 gene), and age (in months) at the time of the acquisition of the radiographs.

For training DL models, we use advanced computers (equipped with the strong GPUs) located within the University of Bonn computing infrastructure.

Ethics

The Bone2Gene project was approved by the ethics commission of the University hospital Bonn on 09.09.2021 (Ref. No. 386/17).

[2] https://www.deeplasia.de

The Team

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- The United States of America
    - National Human Genome Research Institute of the National Institutes of Health (NIH)
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