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“Bone2Gene” Secures €1 Million Grant

The “Bone2Gene” project of researchers from the University of Bonn and University Hospital Bonn has been awarded funding worth €1,000,000 as part of the Federal Ministry of Education and Research’s GO-Bio initial program. The money is now enabling the team to progress to the feasibility phase and get its product ready for market launch. This is actually the second grant that the project has secured, the first having been used for the conceptual phase and to test the product’s marketability. “Bone2Gene” is using artificial intelligence (AI) to make genetic bone conditions known as skeletal dysplasia easier to spot and diagnose.

Harnessing AI to improve the detection of skeletal anomalies

The “Bone2Gene” project was created by two researchers based in the Institute of Genomic Statistics and Bioinformatics at the University Hospital Bonn: Dr. Behnam Javanmardi and Professor Peter Krawitz, who is also a member of the University of Bonn’s Modelling and Life & Health Transdisciplinary Research Areas. After securing their first GO-Bio grant in 2023, they also brought Professor Klaus Mohnike from the Central German Competence Network for Rare Diseases at the University Hospital Magdeburg, on board as a partner. “In our project, we’re using AI to spot genetic and rare bone diseases faster,” Dr. Javanmardi explains. “We’re training it on some of the characteristic patterns that skeletal dysplasia exhibits and thus helping medical professionals make their assessments.” The sheer variety of patterns in the skeleton that have to be identified during diagnosis makes it hard even for experienced clinicians to give a clear verdict, resulting in occasional misdiagnoses and forcing patients to wait several years. However, the hope is that the new software-based AI technology will make this easier.

What makes the technology really unique is its ability to identify several different conditions that come under the umbrella of skeletal dysplasia. Besides a quicker diagnosis, this could also help inform treatment decisions for some diseases. More than 700 different rare skeletal dysplasias can be attributed to genetic defects in over 500 different genes. Specific medications have been approved in recent years and, when diagnosed early, allow for a personalized therapy. “We’ll be using our funding to press on with the groundwork for spinning off Bone2Gene,” Dr. Javanmardi says, outlining the next steps. “To this end, we’re currently on the lookout for some strategic partners for a successful market launch.”

Encouraging innovations from within the University’s walls

“Securing this follow-up grant demonstrates the highly innovative nature and significant market potential of this research-derived project, and we’re looking forward to accompanying the team as they continue on their journey,” adds Sandra Speer, Head of the Transfer Center enaCom, which provided detailed advice and support to the project while its proposal was being prepared and submitted. Her team of start-up and innovation consultants works together with the Transfer team in the Faculty of Medicine to support life sciences spin-offs and innovative applications with a research background. Professor Bernd Weber, Dean of the Faculty of Medicine at the University of Bonn and interim CEO of the University Hospital Bonn, highlights: “Technology translation and transfer are particularly important to me in my role as

Dean of the Faculty of Medicine, and we've invested continuously in the quality and scope of our support in recent years. It's fantastic to see the assistance provided by the Faculty's Transfer team deliver its first major successes, and I'm looking forward to continuing to follow and support Bone2Gene's development."

About the GO-Bio funding initiative

The GO-Bio funding initiative is targeted at life sciences researchers with innovative ideas that they will not usually have put into practice yet, which are to be finalized on a conceptual level and turned into an actual application in the two-stage process. The year-long conceptual phase is devoted to formulating ideas, devising implementation strategies and identifying key partners. The projects are then re-evaluated in a second selection procedure, with the successful ones going through to the feasibility phase. Lasting up to two years, this provides an opportunity to demonstrate how the idea could actually work in real life.

Information about Bone2Gene: <https://bone2gene.org/>

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Prof. Dr. Peter Krawitz (left) and Dr. Behnam Javanmardi (center) discuss bone features with Dr. Martha Dohna, child radiology expert at UKB.

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