



## AN ISRAELI SME DEVELOPING AI-BASED INTERPRETATION ENGINE FOR GENOMIC DATA

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Looking to join a consortium settings up a proposal for submission under IMI call topic 5 - [Shortening the path to Rare Disease diagnosis by using newborn genetic screening and digital technologies](#)

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### ABOUT GENOOX

[Genoox](#) is a Healthcare Technology company developing an AI-based interpretation engine for genomic data. Our engine is built and trained on top of the Genoox Community Data - a unique crowdsourced dataset which differentiates our solution making it accurate and based on real-world evidence. To date, the interpretation engine supports multiple applications including rare diseases, oncology, hereditary cancer and carrier screening.

**We are looking to join a consortium, where we would take the lead or active role in AI and novel algorithm development and data analysis.**

### The consortia that may benefit from our contribution, experience and expertise:

An established or forming consortia that have partners with access to patients and clinical data, high familiarity with RD resources and proven experience in developing and validating genetic tests.

### Our specific potential contribution:

Our role in a potential consortium could be leading and achieving **objective 2**: “Federation of available RD databases into a RD metadata repository amenable to machine learning or other advanced digital tools”; which is an area we are highly experienced in, and have already established significant assets. **The Genoox Platform already assimilates data from millions of data points in order to create a federation of databases, from public data sources, private, community repositories and BioBanks. On top of these aggregated databases, we apply machine-learning algorithms in order to have an automated variant classification, prioritize genetic data according to its clinical background and suggest evidence, which was found in similar cases and might have relevance for current cases.**

Another object we can lead is **objective 4**: “Repurposing of pre-existing diagnosis AI algorithm to identify early onset RD patients in electronic health records (EHRs) and Design and development of new AI algorithm(s) to achieve the above goal”. **We have developed an AI based NLP engine which extracts evidence from publications, by detecting both genetic data and phenotypic terms from over 30 millions scientific publications. In addition, our NLP algorithms are used today to extract structured data such as HPO terms from unstructured data such as text blurbs and EHR data.**

**The Genoox platform is being used by hundreds of organizations around the world for solving rare disease cases based on whole exomes and whole genomes.** Our team is composed from highly skilled bioinformaticians, data scientists, software engineers as well as geneticists and clinicians. Our community edition is freely available at <https://franklin.genoox.com/> and can give a sense of what we do and excel at. To date we won multiple grants from the Israeli Innovation Authority and [BIRD foundation](#) and are experienced in working and leading similar projects. We were [recognized](#) as one of top 5 startups disrupting healthcare with AI by Business Insider and Customer Value Leadership award for Genetic Analysis Market by Frost & Sullivan.

Groups that are setting up a proposal are invited to contact:

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