

Publication on Hereditary Angioedema Kininogen Assay (HAEKA) Study

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/EINPresswire.com/ -- A research paper on the frequently misdiagnosed disease Hereditary Angioedema (HAE) and the currently running clinical study 'Hereditary Angioedema Kininogen Assay (HAEKA)' was recently published in the international Orphanet Journal of Rare Diseases.

Patients with HAE are characterized by unpredictable and painful attacks of swellings on different parts of the body. The HAEKA study aims to identify

prognostic biomarkers for HAE – molecules that reflect the disease status and are able to predict attacks before they physically start. This would improve patient's life considerably as they can observe the biomarker level, take immediate actions, and prevent attacks. In the HAEKA study, participants receive study boxes including lancets for finger pricking that allows self-sampling of blood directly during swellings. Such a study design was never applied before for HAE research and gives them hope for exciting discoveries.

The HAEKA study is a unique research collaboration between industry and academia. CENTOGENE GmbH is organizing the study, whereas the Charite is the most important enrolling study center, and Takeda sponsors the study. The concept of the HAEKA study was developed by Prof. Arndt Rolfs, Founder and Ex-CEO of CENTOGENE, Founder of [Arcensus](#), together with Prof. Marcus Maurer and Prof. Markus Magerl from Charite. Both Prof. Rolfs, as well as the first author Dr. Toni Förster (Operational Project lead) and the last author Dr. Volha Skrahina (Director Clinical Studies) are working by now for Arcensus GmbH, a medical start-up company for commercial genetic diagnosis via Whole Genome Sequencing (WGS).

As the study is still ongoing, no final results are available yet. However, the paper presents the current status and intermediate data of the HAEKA study. It further raises awareness both about the disease HAE (for publicity) and about the study per se (for interested HAE patients). Importantly, this paper is a creative example of how to find solutions for operating clinical



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studies during COVID-19. The idea of self-sampling from home provides not only flexibility to participants, but also depicts the only way of dealing with the strict contact restrictions.

The publication is available with open access:

<https://ojrd.biomedcentral.com/articles/10.1186/s13023-021-02021-x>

About Hereditary Angioedema (HAE)

Hereditary Angioedema is a frequently misdiagnosed disease, caused by a genetic defect. Patients with HAE develop recurrent swellings (edemas) of various body parts. The edemas are triggered by certain factors like stress or physical stimuli, but they are still unpredictable. They can be even life-threatening if edemas are blocking the larynx. Efficient medication to prevent or ameliorate swellings are available, however prognostic biomarkers to find the right medication, dosage, and time of treatment are required to improve the lifestyle of HAE patients.

About Arcensus

Arcensus GmbH offers sophisticated genomics testing and preventive health services to give all individuals certainty and actionable insights about their health. The basis of the knowledge-generating direct-to-consumer business is an innovative WGS strategy. WGS captures all genetic variants that are missed in a single gene or panel testing. The direct-to-consumer business model allows the early identification of individuals suffering from a potential genetic disorder or those at risk for such problems. Such a strategy is particularly valuable when diseases are life-threatening or even fatal as sudden death or aortic dissection at their first presentation.

From our headquarters in Rostock and Berlin, Germany, a cross-functional team of genetic experts, medical doctors, and data scientists works to make the most sophisticated and best genetic interpretation with medical and health prevention applications accessible to everyone. For more information, visit <https://arcensus-diagnostics.com/>

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