Scientists develop diagnostic tool for Familial Mediterranean Fever

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Researchers at VIB and Ghent University have developed a tool to diagnose Familial Mediterranean Fever (FMF). Particularly common among Mediterranean populations, this genetic disease is characterized by inflammation, fever and severe pain. Because of its complex diagnosis, patients often remain untreated for many years, which can eventually lead to kidney failure. In collaboration with Ghent University Hospital and Antwerp University Hospital, VIB and Ghent University are now planning clinical trials to further validate immunodiagnosis of FMF. The study is published in the leading scientific journal *PNAS*.

In the Mediterranean basin, including the Middle East and Caucasus, FMF has a prevalence between 1 and 2 per 1,000 inhabitants. FMF is usually diagnosed during childhood, after which a daily, lifelong treatment is necessary. However, accurate diagnosis is complicated by a number of factors: other autoinflammatory diseases show similar symptoms, the clinical picture is often incomplete in young children, atypical signs may occur, and a suggestive family history is sometimes lacking. Wrong or late diagnosis often even leads to unnecessary surgery and, ultimately, kidney failure.

The lab of professor Mohamed Lamkanfi (VIB-Ghent University) developed an alternative for today's inadequate diagnosis, efficiently segregating FMF patients from people suffering from other autoinflammatory diseases and healthy individuals. The tool detects changes in the body's immune reaction to pyrin, a protein that is usually mutated in FMF. Following successful tests on mice, the tool has been

validated in 13 patients in collaboration with physicians from Belgium and Italy.

Prof. Mohamed Lamkanfi (VIB-Ghent University): "As next steps, we are setting up clinical trials in Belgium for which we are actively seeking volunteers – both FMF patients and people suffering from related inflammatory disorders. These trials are funded by, among other parties, the European Research Council and FWO (Research Foundation – Flanders). In addition, labs from the Netherlands and Italy have already expressed interest. We are also exploring possible collaborations with industrial partners in order to make our method available as a diagnostic kit."

The new tool could be good news for thousands of people with a Mediterranean background. However, in rare cases, FMF also affects other people. Prof. Lamkanfi and his team have expressed the ambition to start the clinical trials as soon as possible. Meanwhile, a patent application for the invention has been submitted.

More information: Hanne Van Gorp et al. Familial Mediterranean fever mutations lift the obligatory requirement for microtubules in Pyrin inflammasome activation, *Proceedings of the National Academy of Sciences* (2016). DOI: 10.1073/pnas.1613156113

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