Precision medicine new opportunity for complex diseases

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A new review article looks at readiness prior to the implementation of genomics-based precision medicine in complex diseases. The extensive work has been led by researchers at Karolinska Institutet and Lund University and has been carried out together with some 30 researchers throughout Sweden. The results are now being published in the *Journal of Internal Medicine*.

"Complex diseases are the next area where we see that precision medicine can play a significant role in how a large group of patients with different diseases is both diagnosed and treated, but also how we prevent these health scourges," says Richard Rosenquist Brandell, Professor at the Department of Molecular Medicine and Surgery, Karolinska Institutet and Director of Genomic Medicine Sweden.

Complex diseases consist of common health scourges with heredity, environment, and lifestyle affecting the risk of disease. The researchers behind the article see great potential in an increased use of precision medicine based on genomic analyses for these disease groups, which can directly improve and personalize diagnostics, treatment, and prognosis. This report lays the foundation for a national starting point to take the next step and implement existing knowledge in clinical practice and focus on future research initiatives where the needs are greatest.

"An important example where precision medicine is expected to be of great importance is the use of biological drugs in various inflammatory diseases, such as asthma, inflammatory bowel disease (IBD), multiple

sclerosis (MS), and rheumatoid arthritis (RA). Many of these new medicines have a very specific mechanism of action and there is a great need to be able to predict the future course of disease for an individual patient and to see how they respond to treatment with different drugs, i.e. to choose the right medicine for the right patient," says Erik Melén, Professor at the Department of Clinical Science and Education, Södersjukhuset, Karolinska Institutet, one of the main authors of the article.

Another example is when genomic analyses are used to rule out a serious genetic disease in patients that show a clinical picture very similar to a complex disease. This information directly affects treatment and prognosis and how the patient is to be followed up.

"One of my patients, a six-month-old boy, had severe asthma-like conditions, and we couldn't determine whether it was an early-onset form of severe asthma or if it was a congenital disease. We did whole-genome sequencing to look at several genes that could be involved and we could then rule out a congenital genetic disease," says Erik Melén. Genomic analyses are already an important tool for working with patients who have a more difficult form of complex diseases.

For some complex diseases, clinics today are ready to start using genomics-based precision medicine in clinical practice. For the vast majority, the next step is to fill in the knowledge gaps that still exist. For example, by adding genomics as part of clinical trials to further optimize diagnostics, treatment choices, and treatment efficacy in different studies.

More information: P. W. Franks et al, Technological readiness and implementation of genomic-driven precision medicine for complex diseases, *Journal of Internal Medicine* (2021). DOI: 10.1111/joim.13330

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