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Whole-genome sequencing improves diagnosis of broader range of genetic diseases

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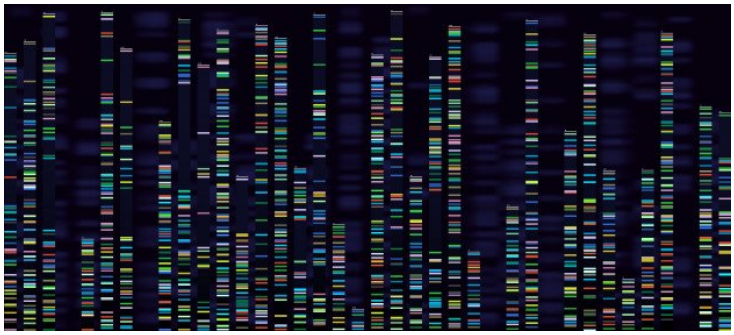
Illumina and the University Hospital of Tübingen assess the value of whole-genome sequencing (WGS) as a first-line diagnostic test for patients with genetic diseases and familial cancer syndromes



Illumina, Inc. on Feb 19, 2021 announced an agreement with the Institute of Medical Genetics and Applied Genomics at the University Hospital of Tübingen to assess the value of whole-genome sequencing (WGS) as a first-line diagnostic test for patients with genetic diseases and familial cancer syndromes. Illumina will support the new investigator-initiated study, called the Ge-Med Project, with sequencing, analysis and health economic expertise.

The Institute is the first laboratory in Germany accredited to perform clinical WGS. Previously, it used whole exome sequencing for the diagnosis of rare disease conditions which involves sequencing only around 1% of the genome known to contain the coding regions that provide instructions for making proteins.

The move to WGS is based on a two-year feasibility study by the Institute, supported by Illumina, which found that WGS provided improved diagnosis across a range of rare diseases. For example, as many as 75% of genetic eye diseases were accurately diagnosed using WGS, including some forms of disease that could only be identified by sequencing the entire genome. Similar results were found for rare childhood cancers and for conditions that cause developmental delay in children.

In addition to expanding the range of conditions for diagnosis, the new study will examine the ability of WGS to generate scores for the risk of common diseases based on genomic data. Known as a polygenic risk score, this will help identify individuals that may benefit from personalized healthcare management.