CeGaT Brings New Dimension to Exome Diagnostics

CeGaT aims to solve all genetically-caused cases of disease to help physicians state a diagnosis. To pursue this goal, CeGaT incorporated its long-term expertise and latest scientific knowledge in an innovative diagnostic tool: CeGaT Exome Xtra is the most powerful genetic diagnostic option for patients with complex, unspecific, and rare diseases.

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CeGaT has been utilizing the power of genetic diagnostics for more than a decade as it introduced gene panels to clinical genetic diagnostics in 2010. Since then, the company has been continuously updating the panels using the newest literature and in close collaboration with clinical experts. Based on all the knowledge gained, CeGaT Exome Xtra is the latest innovation in genetic diagnostics, particularly suitable for solving the most complex patient cases.

With CeGaT Exome Xtra, patients and their treating physicians benefit from a unique approach that combines the advantages of whole-exome sequencing (WES) and whole-genome sequencing (WGS), while avoiding their disadvantages. It outperforms commercially available WES and WGS and increases diagnostic yield.

CeGaT Exome Xtra – Better than exome, smarter than genome

CeGaT Exome Xtra binds all accomplishments of genetics in one single test.

- Extra smart clinical design

CeGaT Exome Xtra considers all medically-relevant regions throughout the complete genome, while standard WES only comprises the coding regions. All known pathogenic and likely pathogenic intronic variants are included, as well as disease-associated individual transcripts and cryptic exons. Regular updates ensure that it always corresponds to the latest science and thus provides the ideal basis for genetic diagnostics.

- Extra thorough analysis

CeGaT's data analysis goes beyond regular exome diagnostics as it accounts for variants in genes with reduced penetrance, variable expressivity, and imprinting effects. It furthermore assesses copy number variants (CNVs), also including combinations of sequence variants (SNVs, InDels) with CNVs. All family constellations (duo, trio, etc.) can be analyzed.

- Extra insightful results

A total of four scientific experts and medical doctors contributes to every medical report – generating the best possible results with maximum medical usability.

CeGaT Exome Xtra outperforms WGS in a diagnostic setting.

Although WGS is sometimes described as the most comprehensive genetic analysis possible, its coverage is often too low for reliable variant detection, and mosaicism cannot be detected. CeGaT Exome Xtra covers more relevant regions at higher coverage and thereby delivers higher sensitivity – at much lower costs than WGS. At the same time, thousands of irrelevant variants usually obtained by WGS analyses are avoided, improving diagnostic speed and accuracy.

Whole-genome sequencing is a great tool for research. Until it is ready for diagnostic use, CeGaT Exome Xtra provides the clinical genome diagnostics to help patients today.

All exome analyses include the regular re-evaluation variants of uncertain clinical significance (VUS) for two years to further increase the diagnostic yield. This service is free of charge.

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Further information

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