



BIOSCIENTIA
INTERNATIONAL

THE NEXT CHAPTER OF DNA ANALYSIS

TRANSLATING EXOME SEQUENCING INTO CLINICAL BENEFIT



// MORE THAN 20,000 PAGES AND UP TO 50 MILLION LETTERS ...

make up the exome chapter



This constitutes less than two per cent of the entire genome. But it is the part of the human genetic material which contains 85 per cent of the known disease causing mutations.

This plethora of information will help us to get a better understanding of the mechanisms underlying diseases.

In order to obtain meaningful results, the technical quality of sequence data is as important as the quality of the bioinformatic data analysis to enable the best medical interpretation of genetic variants.

Complex, unspecific symptoms?
Uncertain diagnosis?

In these cases, exome sequencing is the method of choice to

- pinpoint the disease cause,
- determine appropriate treatment,
- analyse the risks of recurrence or the probability of patients or their relatives to develop a disease.

THE BENEFITS OF OUR EXOME DIAGNOSTICS AT A GLANCE:

- **Flexible**
Exome data allow us to analyze any group of genes based on your clinical question and interpret these as customized gene panels.
- **Fast**
Shorter processing times owing to our in-house bioinformatics department, efficient and optimized analysis processes and powerful sequencing technologies (NovaSeq 6000).
- **Accurate**
To interpret the numerous variants obtained in an exome analysis rapidly and selectively, we include all information on the clinical picture and the patient's symptoms. This requires interdisciplinary skills. Our specialists that is biologists, bioinformaticists and geneticists work closely together to achieve this.
- **Comprehensive**
We offer trio exome sequencing to interpret or determine de novo mutations with greater accuracy. Together with the patient samples, we analyse the DNA of the parents. This increases the detection rate and is time- and cost-effective



// HOW TO REQUEST:

- Request form including the signed declaration of consent of the patient or his parents
- Sample material: 2 - 5 ml EDTA blood (room temperature).

Would you like to learn more about the potential of exome sequencing? Then call us at +49 6132 781 240 or write an e-mail to info.genetik@bioscientia.de



SPEED, SAFETY AND SERVICE QUALITY

Fast and reliable sample transport due to close proximity to Frankfurt Airport. This means: short ways, fast processing and rapid results.



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