BIOSCIENTIA
INTERNATIONAL

NEXT GENERATION SEQUENCING

REVOLUTIONARY IMPACT FOR DOCTORS, PATIENTS AND FAMILIES

// IT'S ALL ABOUT THE PATIENT

„I had many different diagnoses in the meantime. Including diagnoses that were very misleading. As a result of which I also made some suboptimal choices in my career."

Patients with rare diseases and non-specific symptoms are often examined for years without ever getting a reliable diagnosis. Genetic diagnostics with its innovative NGS approaches can be of decisive help here, through:

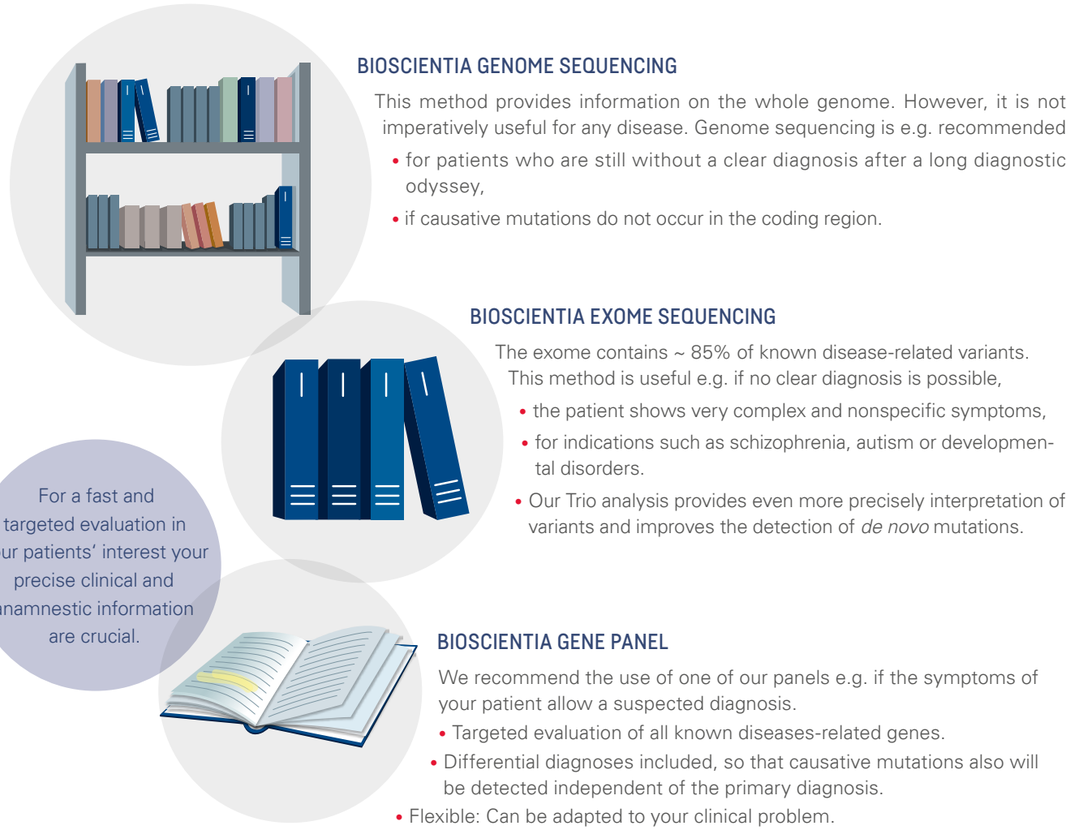
- finding pathogenic mutations quickly and effectively,
- specific information about prognosis, spectrum of disease and recurrence risk for family planning purposes,
- guidance for persons affected („naming the disease"),
- avoiding painful invasive procedures, e.g. in case of muscular dystrophy,
- leading the patient to an – often very specific – therapy.

It is in your power: Cut your patient's diagnostic process short. For the sake of a contemporary and individual treatment, more and more doctors integrate our genetic diagnostics into their practice or clinic routine. Why don't you, too, improve your medical care now by using our support in all questions on analysis and interpretation of reports as well as in diagnostic and therapeutic issues?

For a brief overview of our Next Generation Sequencing approaches see the next page. As every patient situation requires its own approach, our experts are happy to assist you in choosing the best diagnostic strategy in advance.

KEY TO PRECISION MEDICINE

Different needs call for different approaches.



For a fast and targeted evaluation in your patients' interest your precise clinical and anamnestic information are crucial.

BIOSCIENTIA GENOME SEQUENCING

This method provides information on the whole genome. However, it is not imperatively useful for any disease. Genome sequencing is e.g. recommended

- for patients who are still without a clear diagnosis after a long diagnostic odyssey,
- if causative mutations do not occur in the coding region.

BIOSCIENTIA EXOME SEQUENCING

The exome contains ~ 85% of known disease-related variants. This method is useful e.g. if no clear diagnosis is possible,

- the patient shows very complex and nonspecific symptoms,
- for indications such as schizophrenia, autism or developmental disorders.
- Our Trio analysis provides even more precisely interpretation of variants and improves the detection of *de novo* mutations.

BIOSCIENTIA GENE PANEL

We recommend the use of one of our panels e.g. if the symptoms of your patient allow a suspected diagnosis.

- Targeted evaluation of all known diseases-related genes.
- Differential diagnoses included, so that causative mutations also will be detected independent of the primary diagnosis.
- Flexible: Can be adapted to your clinical problem.

THE REPORT MAKES THE DIFFERENCE

Sequencing and comparing the results to public databases is what many modern laboratories can do. From us, you get more:

- Convince yourself of our reliable and meaningful reports. In view of the fact that we always take your point of view when evaluating and interpreting results, you will get a reliable diagnosis and your patient management will be supplemented in a profitable manner, thus enabling specific therapies.
- Keep your patients' safety in mind. We work in compliance with the highest quality standards. This is confirmed by the accreditations of national and internationally accepted organisations (e.g. DAkkS, CAP, CLIA).
- Opt for a stable partner: Being a part of Sonic Healthcare, we take a long-term approach and will also be here for you in the future.
- On top: Benefit from our full service that integrates into your practice or clinic routine smoothly and conveniently. For Bioscientia, the analysis request does not end with the report.
  Contact: aerzte-humangenetik@bioscientia.de

// HOW TO REQUEST:

- Fill in the „Molecular genetic diagnosis" request form, incl. declaration of consent.
  Important: We cannot start a diagnosis without a signed consent of your patient.
- Provide 2 to 5 ml EDTA blood (at room temperature).
- Give a suspected diagnosis and anamnestic information.

Download at:
bioscientia.de/genetik/en/methods/gene-panels

We are looking forward to good cooperation.

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INTERNATIONAL

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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