CGC genetics
a Unilabs company

Genetic Testing

Information for health professionals

WHY CHOOSE CGC GENETICS

EXPERIENCE

Since 1992, CGC Genetics is a reference laboratory for genetic diagnostic testing.

GLOBAL SOLUTIONS IN GENETICS

More than 4 000 genetic tests available for all medical specialties, all performed with state-of-the-art technology.

CLINICAL INTEGRATION

The clinical department has 7 Medical Genetics Specialists teaming up with over 60 highly qualified Geneticists working together analyzing and interpreting each case, providing results with a strong clinical integration.

RESEARCH AND DEVELOPMENT

Large investment in R&D for new and/or unique approaches and tests, positions CGC Genetics as a reference laboratory for Medical Genetics Diagnostic Services.

INTERNATIONAL REFERENCE

CGC Genetics is a leading laboratory in Medical Genetics in Europe, receiving samples from >60 countries, including hospitals, public and private, medical clinics, insurance companies, universities and research centers.

GUARANTEED QUALITY

A strict Quality policy adopted by the CGC Genetics has resulted in the implementation of ISO 9001:2008 and in obtaining CLIA licensing (Clinical Laboratory Improvement Amendments).

CUSTOMER CARE

Our customer support team provides personalized assistance to all questions: from test selection to result interpretation aid and additional testing options.

WHAT WE OFFER

NEXT GENERATION SEQUENCING – NGS

NGS Next Generation Sequencing (NGS) is a cutting-edge technology for the sequencing analysis. In a single test, a gene, several genes (panel) or the whole exome are analyzed at a fraction of the time and cost than before. However, not all options available on the market offer the same coverage and, therefore, the same diagnostic yield. The coverage represents the percentage of evaluated target regions and the depth of coverage represents the number of times that a specific target region is sequenced.

NGS PANELS

CGC Genetics's NGS panels consists on the simultaneous sequencing of various genes associated with a disease or phenotype.

CGC Genetics has available more than 200 panels for all medical specialties.

All NGS panels available at CGC Genetics are custom-designed by our team and constantly updated to include additional genes, leading to a strong diagnostic yield.

Please find the latest version of our NGS panels at www.cgcgenetics.com

EXOME SEQUENCING

Exome is the set of all exons, encompassing 1-2% of the human genome. Exome Sequencing is a very efficient strategy to study the exons of a patient's genome, unraveling mutations associated with specific disorders or phenotypes.

With this diagnostic strategy, patients can be studied with a significantly reduced turnaround time and cost, when compared to single gene or gene panel sequencing. CGC Genetics has available two Exome Sequencing options, to best fit with patient's individual needs:

- Whole Exome Sequencing (WES), which analyzes the entire exome with sequencing of about 20 000 genes;
- Disease Exome by CGC Genetics, which analyzes >6 000 most clinically-relevant genes.

| WHOLE EXOME SEQUENCING – WES

WES entails the sequencing or "reading" of all genomic coding regions, i.e., the exons. Genetic variants are searched throughout the 20 000 genes that make up the exome.

WES Advantages:

- Maximum diagnostic capability available at this moment
- Clinical phenotype expansion through the identification of new genes with clinical significance
- Detection of new variants not yet described
- Confirmation of possible variants with Sanger Sequencing

WES has an average depth of coverage of 100x, with more than 90% coverage of coding regions.

| DISEASE EXOME BY CGC GENETICS

Disease Exome by CGC Genetics is one of the biggest sequencing panels available which includes approximately 6 000 clinically relevant genes. Disease Exome panel has a particular focus on coding regions (exons) and in flanking regions (splicing sites).

DISEASE EXOME Advantages:

- Targeted at clinically relevant genes
- High diagnostic capability
- Confirmation of possible variants with Sanger Sequencing
- Report with clinical integration and interpretation
- Solving complex diagnostic cases
- Reduced cost compared with whole exome sequencing (WES)
- Requires only the patient sample (no parent sample needed)

INDEX CASE ONLY VS TRIO SEQUENCING

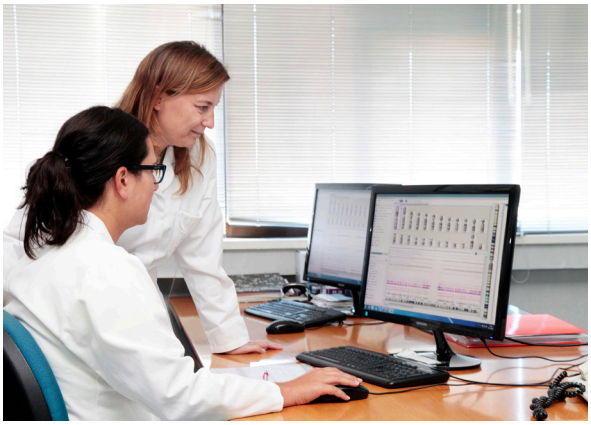
CGC Genetics offers two types of approach for exome sequencing: to analyze only the index case or to analyze both the patient and his/her parents (or other direct family members). Specially for WES, CGC Genetics recommends to perform the latter. This way, it is possible that both parents serve as a reference for filtering benign variants, making Exome Sequencing an more e

ffective diagnostic method in detecting recessive Mendelian diseases and de novo variants.

MITOCHONDRIAL DNA TEST

With exome sequencing, only nuclear DNA is analyzed, excluding mitochondrial DNA. Mitochondrial DNA contains 37 genes involved in several mitochondrial disorders.

CGC Genetics panel for detection of mitochondrial diseases includes the full sequencing analysis of the whole mithocondrial genome by NGS.



ARRAY CGH

Array CGH is a chromosome analysis performed with a microarray technology, i.e., a high resolution technique, which allows a thorough and comprehensive study of the whole genome by identifying copy number variants and events associated with absence of heterozygosity. Currently, its use is recommended as a first tier test to study patients with psychomotor development delay, autism or polymalformative syndromes, and also in prenatal context in high risk pregnancies.

CGC Genetics offers two different resolutions, the highest resolution having the greatest diagnostic capability:

- **Array CGH Cytoscan® 750K (High Resolution)** - 750.000 markers coverage
- **Array CGH Cytoscan® HD (Very High Resolution)** - 2.700.000 markers coverage

Array CGH available at CGC Genetics uses Affymetrix platform, a combined platform which allows to detect, besides DNA losses and gains, absence of heterozygosity (AOH) which is highly important in diagnosis of recessive disorders and in cases of uniparental disomy. This Affymetrix platform used by CGC Genetics is the only FDA-validated platform for use in diagnosis of psychomotor delay, autism and polymalformative syndromes.

CYTOGENETICS

Cytogenetic approaches to detect chromosome alterations and their association to human constitutional and acquired genetic diseases has improved greatly over the past decades. Conventional cytogenetics, including karyotype analysis, was later complemented with fluorescence in situ hybridization (FISH) and comparative genomic hybridization (CGH), and thus enlarging and improving the applicability and reliability of cytogenetic analysis.

CGC Genetics provides a wide variety of cytogenetic tests, using the most advanced technologies available, the most qualified professionals and interpretation of results by our medical geneticists, conferring a well-defined clinical and medical application.

PRENATAL SCREENING

Knowing clinical information about the baby, as quickly as possible, is the aspiration of every pregnant woman.

Nowadays, 1st trimester screening, performed between week 9 and 12 (preferentially at weeks 9 and 10), is considered to be the best prenatal screening approach for Down syndrome (trisomy 21), with high detection rates (90 to 97%) and reduced percentage of false positives rate (3 to 5%). This test includes biochemical parameters (free ßhCG and PAPP-A) and ultrasound markers (nuchal translucency and fetal nasal bone) that are assessed between week 11 and 12. This test also evaluates risk for Trisomy 18 and Trisomy 13.

Please note that a Prenatal Screening with a low risk result does not exclude Down syndrome, Trisomy 18 or other chromosomal alterations, congenital defects, mental retardation or disorders: being a screening, it only indicates a low risk for these conditions.

TOMORROW
non invasive prenatal test

TOMORROW Prenatal Test is CGC Genetics solution for Noninvasive Prenatal Testing. This is a noninvasive technique that aims to investigate the presence of trisomy of chromosomes 21, 18 and 13 in fetal DNA, to identify fetal gender and to detect aneuploidies of sex chromosomes (Monosomy X, XXX, XXY, XYY).

Why TOMORROW Prenatal Test is so important today

EARLY DETECTION
Test can be performed as early as 10 weeks of pregnancy.

SIMPLE
Only a simple blood collection required, with no prior preparation.

SAFE
Performing the test has no risk of abortion.

RELIABLE ANALYSIS
Very low false positive and false negative rates (0.1% and 0.02%, respectively) [1].

TURNAROUND TIME
Report ready in 6 business days on average (max 10)

TWIN PREGNANCY
Detection of trisomy of chromosomes 21, 18 and 13 can also be performed in twin pregnancies (two fetuses).

IVF/EGG DONATION
TOMORROW Prenatal Test can be performed in case of IVF/egg donation (self-donation or not).

NGS TECHNOLOGY
TOMORROW Prenatal Test uses Illumina's Next Generation Sequencing (NGS) technology to study maternal and fetal circulating DNA fragments.

FREE OF CHARGE CONFIRMATION

In case of a positive result, confirmation by prenatal invasive diagnosis is recommended. In this case, CGC Genetics offers confirmation analysis free of charge , by QF-PCR, with results available within 24-48h, as well as chromosomal analysis (karyotype), in a fetal sample.

PLEASE NOTE:
TOMORROW Prenatal Test is not available in USA, Canada, China and Japan.

[1] Taneja et al., Noninvasive prenatal testing in the general obstetric population: clinical performance and counseling considerations in over 85 000 cases. Prenatal Diagnosis. 2016, 36: 1–7. DOI: 10.1002/pd.4766.

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