Unravel the Exome

One high-quality workflow.
Multiple solutions.

Whole Exome Sequencing

Advancements in next-generation sequencing technology have made the use of whole exome sequencing (WES) an affordable option in Genomics research. Implemented in environments that are quality controlled, WES can boost discovery type of studies that generate data with great/immediate translational power.

In clinical settings, WES has transformative potential because it drastically increases diagnostic yields in rare disease cases, improving patient outcomes and reducing health care costs. When combined with effective computational processing and virtual panel design, WES can deliver tailored clinical outputs in a standardised and scalable manner.

Exome sequencing as a first line test:

Exome sequencing captures only the coding regions of the genome. This enables an exome solution to support several applications, including rare and severe genetic conditions, cancer screening and disease risk assessment.

A focus on what matters:

Limiting sequencing to only coding regions allows for cost effective testing without significant loss to diagnostic power. Customized virtual gene panels refine the process further through standardization and automation.

Sensitivity:

Using the xGen® Exome Research Panel from IDT and Illumina next-generation sequencing technology in conjunction with the DRAGEN BioIT Platform for data analysis allows for world class performance. Single Nucleotide Variants are detected with a sensitivity of 99.8% and heterozygous indel detection sensitivity is 97%.

Diagnostic yield:

Through Fabric Genomics, Artisan Biomed connects to the latest in data resources for assigning pathogenicity; also, curates and maintains a local, region-specific data repository.

Results when you need them:

Routine diagnostic exome referrals are completed within 7 weeks and 5 weeks for urgent referral.

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WES	KEY FEATURES
GENES	- 19 396 genes targeted - All protein-coding regions and at least 2 bp into the intronic regions - Mitochondrial DNA genes not covered
COVERAGE	- Highly uniform coverage of the whole exome - Mean target coverage ~75X - ≥94% of targeted bases covered at ≥20X
VARIANTS	- Detects SNV's and INDELS (>30bp) - Variant call sensitivity = 99.76% - Confirmation of Pathogenic and likely Pathogenic SNV's & INDELS by Sanger sequencing

PRODUCT	DESCRIPTION	*PRICE (ex VAT)
WES-R	Research whole exome	R6 000
WES-C	Solo whole exome with clinical interpretation	R10 000
WES-Cx3	Trio whole exome with clinical interpretation	R20 000
Health Screen	59-disease risk exome based gene panel, with clinical interpretation	R8 000
Hereditary Cancer Screen	55-gene, exome based hereditary cancer risk panel, with clinical interpretation	R8 000
WES-C (+)	Whole exome combined with high-resolution aCGH	R14 000
Cyto-Pre	Pre-natal aCGH, using Cytoscan® Optima	R5 200
Cyto-Post	Post-natal aCGH, using Cytoscan® 750k	R7 500
'Custom Panel'	Whole exome based, tailored gene panels	Enquire

**Pricing for options included here valid until 30th Nov 2019*

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PRODUCT FEATURES:

1. Platform: NextSeq 500/550 for all exome applications; Affymetrix GS 3000 for all aCGH applications (array Comparative Genomic Hybridization)
2. Molecular solution: IDT xGen® Exome Research Panel v1.0; CytoScan® 750K and Optima arrays.
3. Coverage: 30X for research, 60X for all clinical applications.
4. Primary data analysis: For exomes, scalable DRAGEN BioIT platform (can process 1000 exomes in 25 hours); for aCGH applications, Chromosome Analysis Suite (ChAS).
5. Variant interpretation: For exomes, Fabric Enterprise 6.1; SNVs and CNVs included.
6. Copy Number Variations (CNVs): The assay detects CNVs within the limitations of the short-read sequencing technology used.
7. Reporting: Modified after standard operating procedures for clinically relevant variants developed through the UK 100,000 Genomes Project (https://www.fabricgenomics.com/app/uploads/2018/07/FG_Poster_GeL_v5.pdf). pathogenic and likely pathogenic variants reported; for research, raw data, BAM file, and vcf data available.
8. Turnaround time (TAT): 7 weeks for all exome based applications, from receipt of sample; 8 weeks for exome/aCGH combis and post-natal aCGH applications; 4 weeks for pre- and post-natal aCGH applications.
9. Process quality control: RM 8398 reference DNA (NIST) used on a continuous basis for internal quality control.
10. Permissible sample input: 1x EDTA blood tube for all applications; gDNA (>200ng total, ≥ 20ng/ul concentration in 1X TE buffer, A260/280 ≥ 1.8, A260/230 ≥ 2.0 for research purposes only.
11. Confirmation: For clinical cases all pathogenic and likely pathogenic variants confirmed by orthogonal methods.
12. Ethics: Ethics number required to register a research project.
13. Clinical forms: One form required for exome applications; one for cytogenetic applications; both include data consent form.
14. Consultation: Consultation will be offered on Thursdays and Fridays, between 9 am and 12 pm SAST in 30-minute intervals; all meetings can be conducted in secure (encrypted) virtual meeting rooms, using MS Teams. Consultations will be free of charge. For bookings please contact exomes@artisanbiomed.co.za (clinical) and exomes@cpgr.org.za (research).
15. Webinars: a 1-hour webinar will be offered once a month, on the last Friday of each month from 12 pm to 2 pm, SAST. For interest please contact exomes@artisanbiomed.co.za.
16. Genetic counselling: Available on request.
17. Information: For research purposes, exomes@cpgr.org.za; for clinical purposes, exomes@artisanbiomed.co.za.
18. Data storage: With consent, in a locally hosted and secure MS Azure cloud environment.

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