# Expected results

## DIAGNOSTIC FINDING

- a mutation in the exome that explains the patient's medical condition.
- informs about the risk of recurrence in future pregnancies.

## SECONDARY FINDING#

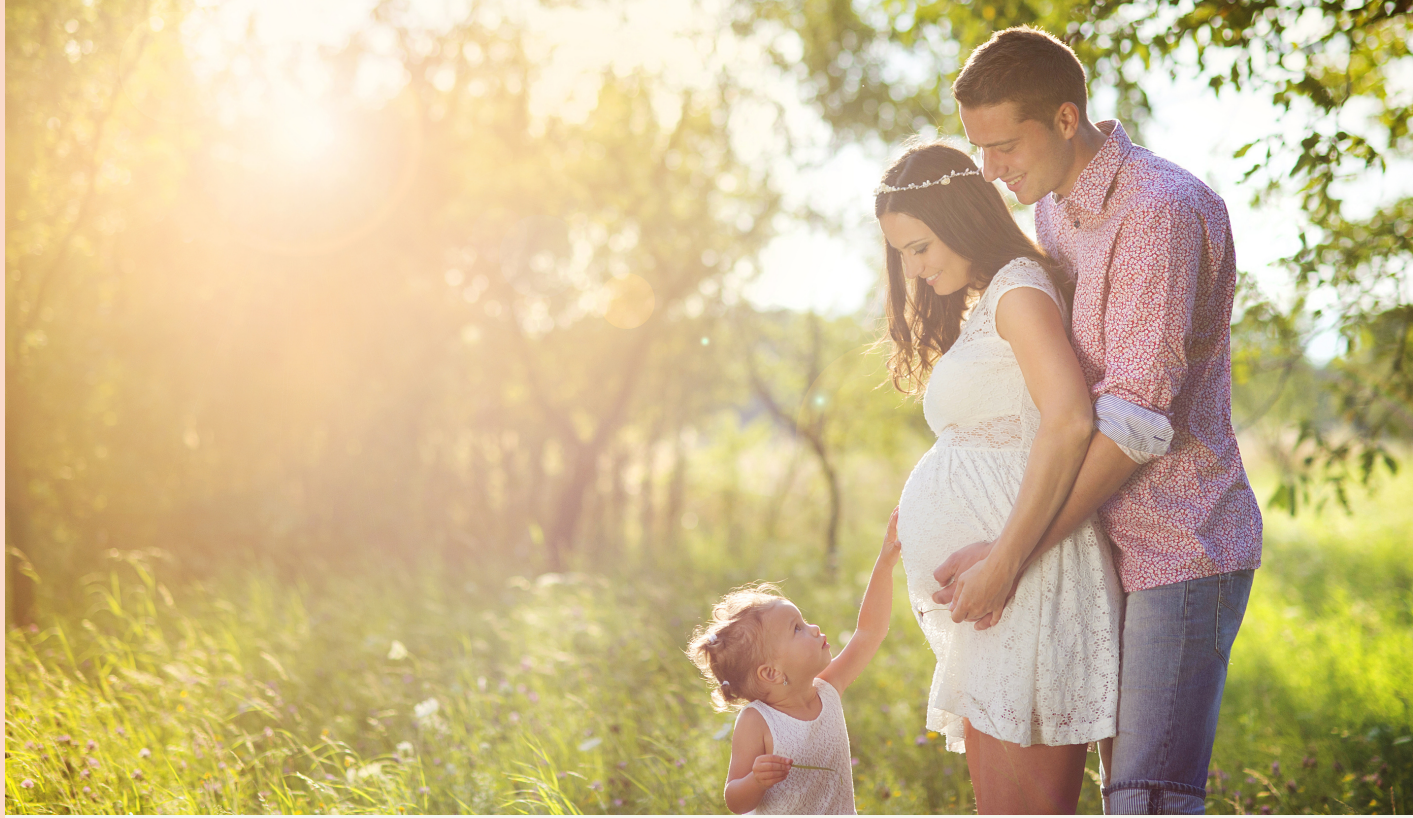
- an unrelated finding to the patient's medical condition.
- may have significant health implications for baby, family, or future pregnancies.

# Opt-in/Opt-out

*Finding an answer to a medical condition through FetalExome may provide life-changing options to treatments and management.*

### Limitations

It is possible a disease-causing variant exists but cannot be characterized using current technologies. Mutations in some genes cannot be detected due to technical reasons including local sequence characteristics or the presence of closely related pseudogenes. Changes from repetitive sequences and repeat expansions are not detected by this method. This analysis is not intended to detect structural variants, copy number variants, variants in mitochondrial DNA, methylation abnormalities, dynamic variants, mosaic/somatic variants. The data analysis and interpretation are performed by the appropriate means of evaluation at the time of testing.



# Contact Us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Provided by CUHK O&G

# FetalExome

## Genetic testing for birth defects and undiagnosed diseases

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Faculty of Medicine
The Chinese University of Hong Kong

# Exome and disease

The human exome constitutes about 2% of the genome which encodes proteins for life function. We know most of the disease causing genetic mutations locate in the exome.

### Human genome: like a book

Human exome is the abstract page

# The test is suitable for

- Isolated anomaly or multiple organ system anomalies

- Suspected monogenic disorder

- Atypical presentation of disease or multiple differential diagnoses

- Undiagnosed cases which exhausting genetic testing options

*Best done as trio pattern, where fetal and both parental samples are timely sequenced and analyzed together.*

# Diagnostic yield

Taking the application of exome sequencing in undiagnosed fetuses as examples*:

- Multisystem anomalies: 31%-33%
- Isolated echogenic kidneys (72%)
- Isolated skeletal (53%)
- neuromuscular/FADS (37%)
- isolated agenesis of the corpus callosum (29%)
- increased NT plus other anomaly at presentation or later (26%)
- isolated hydrops/edema (22%)
- Isolated cardiac abnormalities (11%)

*PMID: 35583085

# FetalExome

### High coverage of most exome

Based on genome sequencing and analysis of the exome, covering nearly the entire protein-coding region of the genome.

### Analyzing all annotated genes

Analysis of the coding regions of more than 20,000 annotated genes for pathogenic or likely pathogenic variants related to patients' clinical phenotype(s).

### Enhancing the detection scope

One-stop detection of single nucleotide variants (SNVs), small insertions and deletions (InDels) and genome-wide absence of heterozygosity at 5Mb.

### Flexibility to know more

Opt in to know your carrier statuses for autosomal recessive conditions and risk for medically actionable diseases. You may also upgrade your test to GenomSeq for comprehensively identifies SNVs, InDels, copy-number variants (CNVs), structural rearrangements (SVs), and absence of heterozygosity (AOHs).

### Quick and responsive service

From sample collection to report issuing: 14 calendar days for trio; 28 canledar days for proband only. Sample types: amniotic fluid, blood, saliva, tissue and DNA.