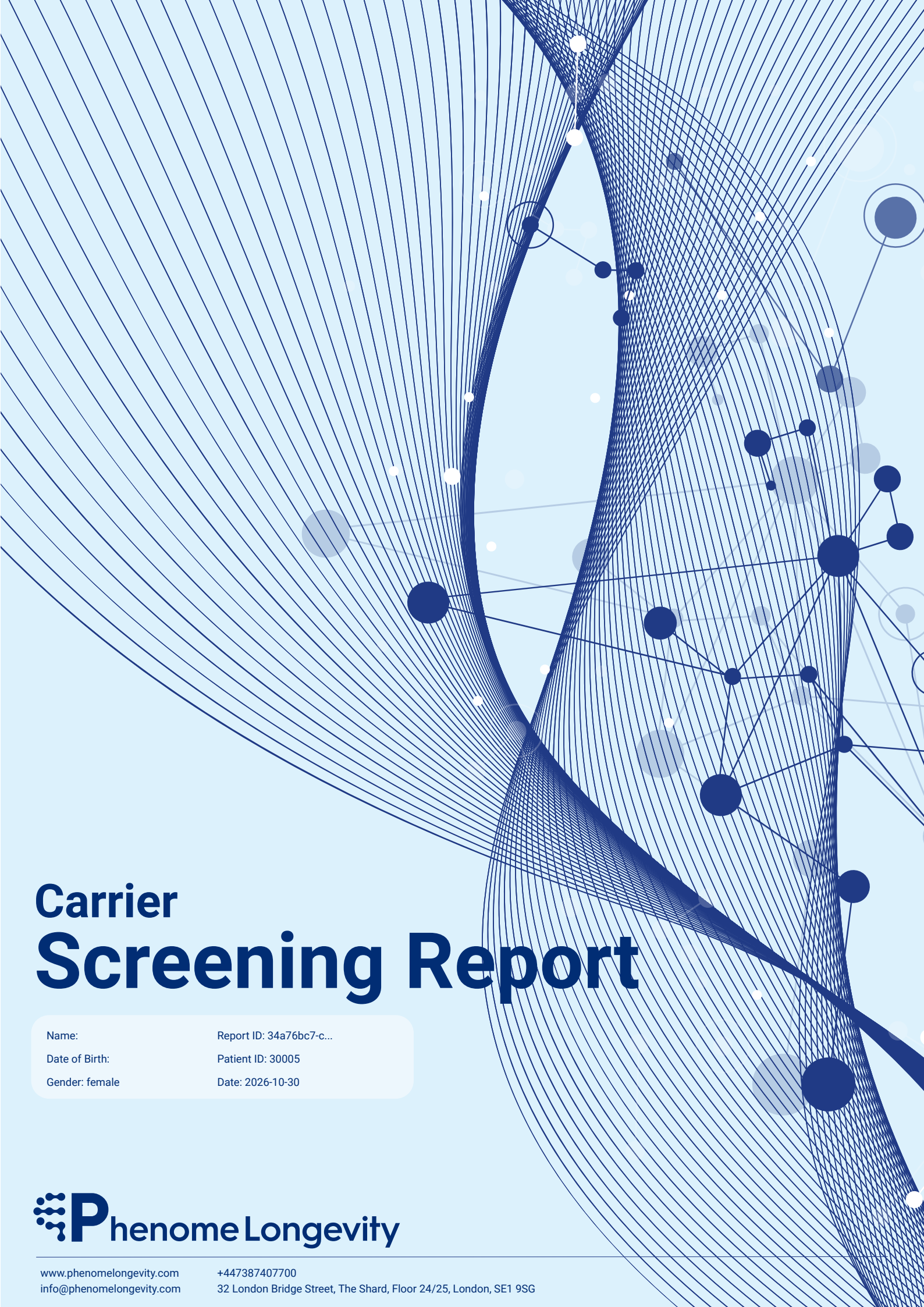


Carrier Screening Report

Name:	Report ID: 34a76bc7-c...
Date of Birth:	Patient ID: 30005
Gender: female	Date: 2026-10-30

Phenome Longevity

www.phenomelongevity.com
info@phenomelongevity.com
+447387407700
32 London Bridge Street, The Shard, Floor 24/25, London, SE1 9SG

+447387407700 info@phenomelongevity.com www.phenomelongevity.com

About Carrier Screening

WHAT IS CARRIER SCREENING?

Carrier screening is a type of genetic test that determines whether an individual carries a copy of a gene variant associated with a recessive genetic disorder. Carriers typically do not show symptoms of the condition, but if both parents are carriers of a variant in the same gene, each pregnancy carries a 25% chance of producing an affected child. This test uses next-generation sequencing (NGS) to analyze hundreds of genes associated with autosomal recessive and X-linked conditions.

WHY IS IT IMPORTANT?

Most carriers are unaware of their status because they do not show symptoms. However, if both partners carry variants in the same gene, their children may be at risk. Carrier screening empowers you with knowledge to make informed reproductive decisions. It can identify risks before pregnancy, guide family planning, and enable access to options such as preimplantation genetic testing (PGT). The ACMG recommends carrier screening for all individuals of reproductive age.

HOW TO READ THIS REPORT

1. **Review Your Results Summary**
   The Introduction page provides an overview of whether any pathogenic variants were detected. A clear result means no actionable variants were found in the genes tested.
2. **Understand Each Finding**
   If variants were detected, each is presented with the disease name, affected gene, inheritance pattern, zygosity, and clinical significance according to ACMG and ClinVar.
3. **Check the Interpretation Guide**
   The Summary page contains definitions for all clinical terms used in this report, including pathogenicity classifications and inheritance patterns.
4. **Consult a Genetic Counselor**
   We strongly recommend discussing your results with a qualified genetic counselor who can provide personalized guidance based on your findings and family history.

KEY TERMS

Pathogenic Variant
A DNA change known to cause or strongly associated with a genetic condition.

Carrier
A person who has one copy of a recessive variant and typically shows no symptoms.

Zygosity
Whether the variant is in one copy (heterozygous) or both copies (homozygous) of the gene.

Inheritance Pattern
How a condition is passed from parents to children (AD, AR, or X-linked).

ACMG Classification
A standard 5-tier system for classifying genetic variants from Benign to Pathogenic.

ClinVar
A public database by NCBI containing reports on the clinical significance of genetic variants.

NOTE FOR CARRIER SCREENING

This report identifies carriers of recessive conditions. A positive carrier result does not mean you have or will develop the condition. It means you carry one copy of a variant that, if also present in your partner, could result in an affected child. Partner testing is recommended.

ABOUT THE TEST Carrier screening is a genomic test that identifies whether an individual carries a pathogenic variant for a recessive genetic condition. Carriers typically do not show symptoms but may pass the variant to their children. This report analyzes clinically relevant genes using next-generation sequencing (NGS) technology.

PATIENT
- **NAME**
- **DATE OF BIRTH**
- **GENDER** Female
- **REPORT ID** 34a76bc7...
- **REPORT DATE** 2026-10-30

PHYSICIAN
- **FACILITY** Phenome Longevity
- **ORDERING PHYSICIAN** Not Specified
- **CONTACT** +447387407700

SPECIMEN
- **SPECIMEN TYPE** Genomic DNA (Blood/Saliva)
- **SPECIMEN ID** 30005
- **ANALYSIS DATE** 2026-10-30

Result Overview

2 Pathogenic Variant(s) Detected

SCREENING MODE Carrier Screening

ANALYSIS Next-Generation Sequencing (NGS)

CONDITIONS IDENTIFIED:

- Biotinidase deficiency
- Congenital adrenal hyperplasia due to 21-hydroxylase deficiency

What This Means

This analysis has identified one or more pathogenic variants in genes associated with recessive genetic conditions. As a carrier, you are generally not expected to show symptoms. However, if your reproductive partner also carries a variant in the same gene, there may be a risk of having an affected child. Genetic counseling is recommended.

GENETIC COUNSELING RECOMMENDED

We recommend discussing these results with a certified genetic counselor for personalized interpretation.

METHODOLOGY

Genomic DNA was extracted and analyzed using a targeted NGS panel covering genes associated with autosomal recessive and X-linked conditions. Pathogenicity was assessed per ACMG/AMP 2015 guidelines and cross-referenced with ClinVar, OMIM, and peer-reviewed literature.

DISCLAIMER

This report is for qualified healthcare professionals and should be interpreted with the patient's clinical and family history. This test does not detect all variant types. A negative result does not exclude genetic conditions.

CARRIER SCREENING — VARIANT RESULTS 2

Biotinidase deficiency 1 / 2

Gene(s)	BTD
Inheritance	AR
Zygosity	Heterozygous
ClinVar Significance	Conflicting
ACMG Classification	Pathogenic

Chromosome	chr3	Position	15645186
Reference Allele	G	Alternate Allele	C
Coding Change (HGVSc)	NM_001370658.1:c.1270G>C	Protein Change (HGVSp)	NP_001357587.1:p.Asp424His
Impact	MODERATE	Max Allele Frequency	0.05533

What This Means

Autosomal recessive: two copies of the variant are needed to cause the condition. Carriers have one copy and are typically unaffected.

One copy of the variant is present. For recessive conditions, this individual is a carrier and typically unaffected.

ClinVar reports conflicting interpretations for this variant. Different laboratories have classified it differently, which means its clinical significance is uncertain. Further evidence or expert review may be needed to clarify its role in disease.

Classified as pathogenic per ACMG/AMP 2015 guidelines, indicating strong evidence that this variant causes disease.

Congenital adrenal hyperplasia due to 21-hydroxylase deficiency 2 / 2

Gene(s)	CYP21A2
Inheritance	AR
Zygosity	Heterozygous
ClinVar Significance	Conflicting
ACMG Classification	Pathogenic

Chromosome	chr6	Position	32039081
Reference Allele	C	Alternate Allele	G
Coding Change (HGVSc)	NM_000500.9:c.293-13C>G	Impact	LOW
Max Allele Frequency	0.004614		

What This Means

Autosomal recessive: two copies of the variant are needed to cause the condition. Carriers have one copy and are typically unaffected.

One copy of the variant is present. For recessive conditions, this individual is a carrier and typically unaffected.

ClinVar reports conflicting interpretations for this variant. Different laboratories have classified it differently, which means its clinical significance is uncertain. Further evidence or expert review may be needed to clarify its role in disease.

Classified as pathogenic per ACMG/AMP 2015 guidelines, indicating strong evidence that this variant causes disease.

Total: 2 variant(s) identified — Carrier Screening

IMPORTANT NOTES

This carrier screening report identifies pathogenic and likely pathogenic variants in genes associated with autosomal recessive and X-linked conditions. Carrier status means the individual has one copy of a pathogenic variant and is generally unaffected. If both partners carry variants in the same gene, each pregnancy has a 25% chance of an affected child. A negative result reduces but does not eliminate carrier risk. Results should be interpreted with genetic counseling.

METHODOLOGY

Genomic DNA was isolated and enriched using a hybridization-based capture panel. Sequencing was performed on Illumina with paired-end reads. Analysis included BWA-MEM2 alignment, GATK/DeepVariant calling, and VEP/ClinVar/OMIM/gnomAD annotation. Classification followed ACMG/AMP 2015 framework. Only Pathogenic and Likely Pathogenic variants are reported. Sensitivity exceeds 99% for SNVs and small indels.

INTERPRETATION GUIDE

Pathogenic	The variant is known to cause disease based on strong evidence from functional studies and clinical databases.
Likely Pathogenic	Strong evidence supporting a disease-causing role but not yet meeting full criteria for definitive pathogenicity. Treated as actionable.
Variant of Uncertain Significance (VUS)	Insufficient evidence to classify as pathogenic or benign. VUS are not reported in this screening.
Heterozygous	One copy of the variant is present. For recessive conditions, the individual is a carrier.
Homozygous	Two copies of the variant are present. For recessive conditions, this typically indicates an affected individual.
AD (Autosomal Dominant)	A single copy is sufficient to cause the condition. Each child has a 50% chance of inheriting it.
AR (Autosomal Recessive)	Two copies are required to cause the condition. Carriers with one copy are typically unaffected.
XR (X-Linked Recessive)	Variant is on the X chromosome. Males with one copy are typically affected. Females are usually carriers.

LIMITATIONS

This test detects SNVs and small indels in targeted regions. It does not detect: large rearrangements (>20bp), repeat expansions, translocations, epigenetic modifications, mosaicism below threshold, deep intronic variants, or variants in genes not in this panel. CNV analysis not included. This test is not a substitute for diagnostic confirmation.

REFERENCES

Richards S, et al. Genet Med. 2015;17(5):405-424. | Landrum MJ, et al. Nucleic Acids Res. 2020;48(D1):D835-D844. | Karczewski KJ, et al. Nature. 2020;581(7809):434-443. | ACMG Board of Directors. Genet Med. 2012;14(8):759-761.