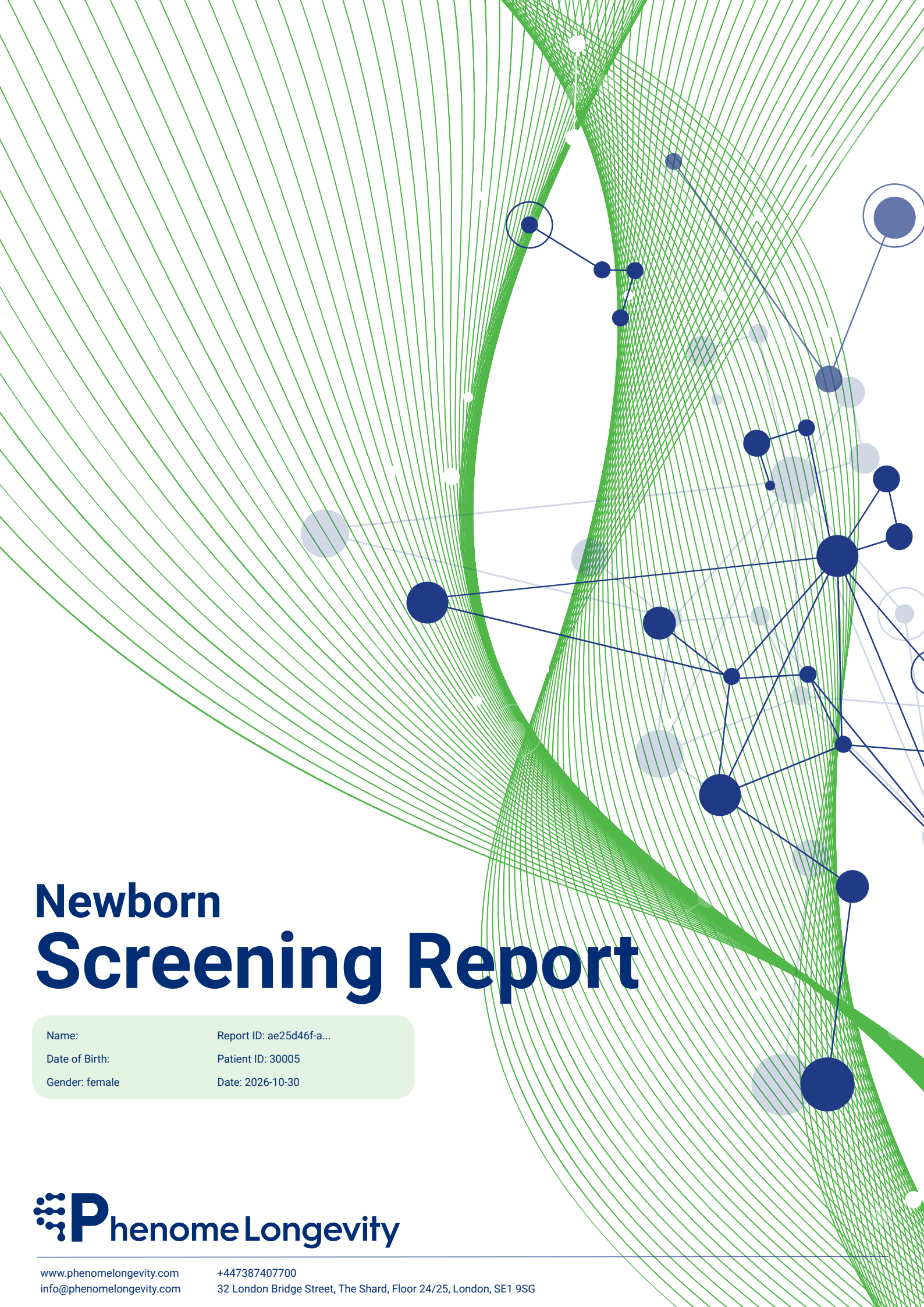

Newborn Screening Report

Name:	Report ID: ae25d46f-a...
Date of Birth:	Patient ID: 30005
Gender: female	Date: 2026-10-30

Phenome Longevity

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About Newborn Screening

WHAT IS NEWBORN SCREENING?

Newborn screening is a comprehensive genomic analysis performed shortly after birth to identify genetic conditions that may benefit from early intervention. Many inherited conditions, if detected early, can be effectively managed or treated to prevent serious health complications. This test uses NGS to analyze genes associated with metabolic, endocrine, hematologic, immunologic, and other treatable inherited disorders.

WHY IS IT IMPORTANT?

Many inherited conditions are treatable when detected early but can cause serious harm if undiagnosed. Genomic newborn screening can identify hundreds of conditions beyond traditional screening. Early detection enables timely treatment, dietary interventions, or specialist care that can dramatically improve your child's quality of life. Some conditions may also have implications for other family members.

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 NEWBORN SCREENING

Positive results should be confirmed through diagnostic testing before clinical decisions are made. Some conditions may require urgent specialist referral. Your healthcare team will guide you through next steps. Not all conditions detected will require immediate treatment.

ABOUT THE TEST Newborn screening is a comprehensive genomic analysis designed to identify pathogenic variants associated with treatable genetic conditions in newborns. Early detection enables timely clinical intervention. This report uses NGS to analyze genes associated with metabolic, endocrine, hematologic, and other inherited disorders.

PATIENT

NAME
DATE OF BIRTH
GENDER Female
REPORT ID ae25d46f...
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 Newborn Screening

ANALYSIS Next-Generation Sequencing (NGS)

CONDITIONS IDENTIFIED:

- Alpha-1-antitrypsin deficiency
- Alpha1-Antitrypsin deficiency

What This Means

This analysis has identified pathogenic variants associated with genetic conditions that may require clinical attention. Early identification enables timely intervention. Findings should be confirmed with diagnostic testing. Specialist referral is strongly 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 comprehensive NGS panel designed for newborn screening, covering genes associated with metabolic, endocrine, hematologic, and other treatable inherited conditions. Classification follows ACMG/AMP 2015 standards.

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.

NEWBORN SCREENING — VARIANT RESULTS 2

Alpha-1-antitrypsin deficiency 1 / 2

Gene(s)	SERPINA1
Inheritance	AR
Zygosity	Heterozygous
Coding Change (HGVSc)	NM_000295.5:c.1177C>T
Protein Change (HGVSp)	NP_000286.3:p.Pro393Ser
ClinVar Significance	Pathogenic/Likely_pathogenic
ACMG Classification	Pathogenic
Impact	MODERATE

Chromosome	chr14	Position	94378529
Reference Allele	G	Alternate Allele	A
Max Allele Frequency	0.001907		

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 both pathogenic and likely pathogenic assertions for this variant across different submitters.

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

This variant has a moderate predicted impact, typically a missense change that alters the amino acid sequence.

Alpha1-Antitrypsin deficiency 2 / 2

Gene(s)	SERPINA1
Inheritance	AR
Zygosity	Heterozygous
Coding Change (HGVSc)	NM_000295.5:c.1177C>T
Protein Change (HGVSp)	NP_000286.3:p.Pro393Ser
ClinVar Significance	Pathogenic/Likely_pathogenic
ACMG Classification	Pathogenic
Impact	MODERATE

Chromosome	chr14	Position	94378529
Reference Allele	G	Alternate Allele	A
Max Allele Frequency	0.001907		

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 both pathogenic and likely pathogenic assertions for this variant across different submitters.

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

This variant has a moderate predicted impact, typically a missense change that alters the amino acid sequence.

Total: 2 variant(s) identified — Newborn Screening

IMPORTANT NOTES

This newborn screening report identifies pathogenic and likely pathogenic variants associated with treatable inherited conditions. Positive findings should be confirmed through diagnostic testing. VUS are not reported. A negative result does not guarantee absence of genetic disease. Clinical monitoring remains essential. Timely referral is recommended for positive findings.

METHODOLOGY

Genomic DNA was isolated and enriched using a capture panel optimized for newborn screening targets. Sequencing on Illumina with paired-end reads. Analysis included BWA-MEM2, GATK/DeepVariant, and VEP/ClinVar/OMIM/gnomAD. Classification followed ACMG/AMP 2015 standards. Only Pathogenic and Likely Pathogenic variants are reported. Sensitivity exceeds 99%.

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.