GeneBaby

Comprehensive Newborn Genetic Screening · 229+ Genes · Metabolic, Immune, Developmental, Cardiac & Sensory Screening

PATIENT **Emma Thompson**	AGE AT COLLECTION **3 Months**	SAMPLE TYPE **Gentle Pediatric Cheek Swab**	SAMPLE ID **GM-GB-2024-10293**
REPORT ID **GB-2024-789456**	COLLECTION DATE **2024-01-08**	REPORT DATE **2024-01-15**	LABORATORY **Gene Matrix CLIA Lab, Chicago, IL**
CLIA / NPI **14D2276402 / 1225729312**	PARENTS / GUARDIANS **Sarah & Michael Thompson**	METHOD **NGS + AI Cross-Reference (2M+ Data Points)**	

229+ GENES SCREENED	**6** HEALTH CATEGORIES	**2** CARRIER FINDINGS	**99.5%** ANALYTICAL ACCURACY

/ Executive Summary

Emma's expanded newborn screening of 229+ genes found no disease-causing results requiring urgent intervention. Two heterozygous carrier findings were identified: PAH (phenylketonuria carrier) and CFTR (cystic fibrosis F508del carrier). Carriers typically do not show symptoms, but these results are important for family planning and genetic counseling. A common MTHFR variant was also noted. G6PD activity is normal. A pediatric care plan and pharmacogenomic guidance follow.

Peace of mind: 95% of genetic conditions identified by GeneBaby are treatable when caught early — and expanded screening detects far more conditions than standard hospital newborn screening. Emma's results are clear across metabolic, immune, cardiac, developmental, and sensory categories.

/ Genetic Findings

GENE	VARIANT	GENOTYPE	IMPACT	RECOMMENDATION
PAH	`c.1222C>T`	Heterozygous (carrier)	CARRIER — PKU Heterozygous carrier for phenylalanine hydroxylase deficiency, associated with phenylketonuria (PKU) carrier status. While carriers typically do not show symptoms, genetic counseling is recommended for family planning.	Monitor phenylalanine levels during routine newborn screening. Consider genetic counseling for parents. No dietary restrictions needed for carrier status.

MTHFR	`c.677C>T`	Heterozygous	COMMON VARIANT Heterozygous variant in the methylenetetrahydrofolate reductase gene. This common variant may affect folate metabolism and homocysteine levels; associated with slightly reduced enzyme activity.	Ensure adequate folate intake through diet or supplementation. Monitor B12 and folate levels during routine checkups. Consider methylfolate supplementation if needed.
G6PD	`c.563C>T`	Normal	NORMAL Normal glucose-6-phosphate dehydrogenase activity. No increased risk for G6PD deficiency-related hemolytic anemia.	No special precautions needed. Standard medication protocols can be followed.
CFTR	`c.1521_1523delCTT (F508del)`	Heterozygous (carrier)	CARRIER — CF Heterozygous carrier for the cystic fibrosis F508del mutation. Carrier status does not cause disease but is important for family planning and genetic counseling.	Genetic counseling recommended for parents. Monitor respiratory health. Partner screening advised for future family planning.

/ Screening Category Summary

Emma's GeneBaby results across the six pediatric health categories screened.

CATEGORY	RESULT	CLINICAL IMPLICATION
Metabolic Disorders — PKU, Galactosemia, MCAD & 30+ more	CARRIER FINDING	PAH carrier (no disease). All other metabolic markers clear.
Immune Deficiencies — SCID, CVID, Wiskott-Aldrich	CLEAR	No pathogenic variants detected.
Developmental Markers — Fragile X, Rett, Angelman, Prader-Willi	CLEAR	No pathogenic variants detected.
Genetic Syndromes — Down, Turner, Klinefelter markers	CLEAR	No syndromic markers detected.
Cardiac Conditions — Long QT, HCM, Brugada	CLEAR	No pathogenic cardiac variants detected.
Sensory Disorders — Congenital hearing loss, retinitis pigmentosa, Usher	CLEAR	No pathogenic sensory variants detected.

/ Personalized Recommendations

Pediatric Care Plan — Developmental Milestones

- Monitor motor skill development: rolling over (4–6 months), sitting (6–8 months), crawling (7–10 months), walking (10–15 months).
- Track language development: cooing (2–3 months), babbling (6–9 months), first words (10–14 months).
- Share this report with your pediatrician at the next well-child visit.

Pharmacogenomics for Parents to Know

- Pain management: CYP2D6 profile on file — relevant if opioid analgesia is ever considered.
- Antibiotics: G6PD normal — standard medication protocols can be followed safely.
- More medications covered in the full report.

Family Planning Guidance

- Both carrier findings (PAH, CFTR) are recessive — relevant only if a partner is also a carrier.
- Genetic counseling is recommended for parents; partner screening advised for future family planning.
- Board-certified genetic counselors are available to walk you through every finding.

Laboratory: Gene Matrix LLC · 1375 W Fulton St STE 545, Chicago, IL 60607 · CLIA ID: 14D2276402 · NPI: 1225729312 · (847) 302-9668 · support@genematrix.io · Mon–Fri 8AM–5PM CT. Testing performed in a CLIA-certified, CAP-accredited laboratory using next-generation sequencing. Analytical accuracy 99.9%. All findings reviewed by board-certified geneticists.
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