BAYLOR GENETICS

Outpatient

Pediatric · Adult

Global MAPS®

(Metabolomic Assisted Pathway Screen)

Comprehensive Metabolomic Testing to Inform Diagnosis of Inborn Errors of Metabolism

Characterize Heritable Metabolic Disorders for Patients with Unexplained Symptoms or Uncertain Genetic Results

For Your Patients Still Searching for Answers, Metabolomics May Make the Difference

Many patients with inherited metabolic disorders, also known as inborn errors of metabolism (IEMs), can present with complex, overlapping metabolic and neurologic symptoms, such as developmental delay intellectual disability, or seizures, making diagnosis challenging.

Routine Biochemical Testing

Targeted testing may require multiple metabolic panels to piece together a full picture.

Genetic Testing

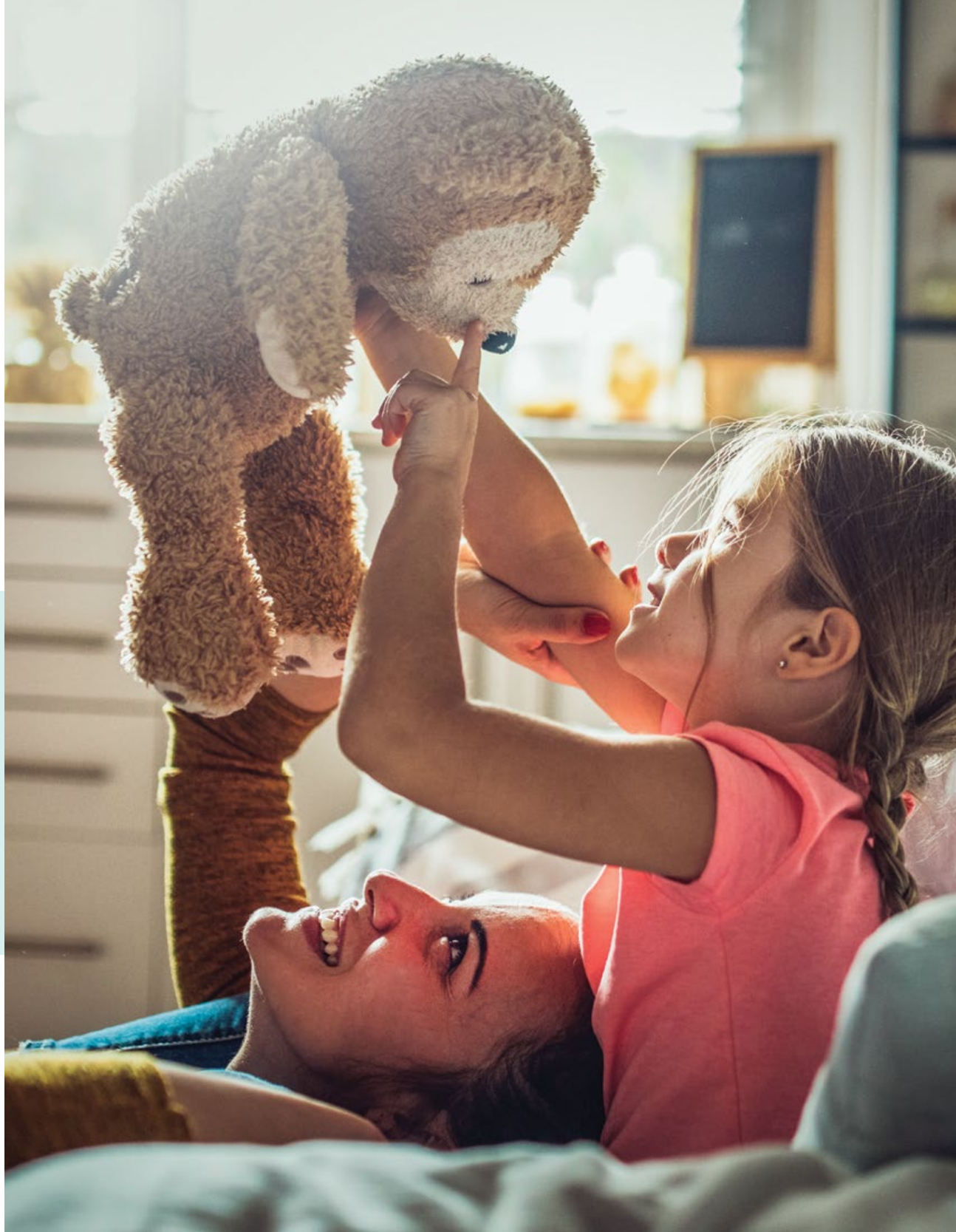
Genetic testing results may not align with or explain clinical presentation.

Early recognition of IEMs can significantly impact a patient's care trajectory, but traditional testing methods often leave gaps that can delay care or miss diagnoses.

1 in 2,500 newborns has an inherited metabolic disorder[1]

Many of these heritable metabolic conditions are treatable, and timely diagnosis can help prevent or reduce health complications.[2] An accurate diagnosis can lead to effective therapies, diets, and even enzyme replacement for some IEMs.[3]

Global Metabolomic Assisted Pathway Screen (Global MAPS®) explores hundreds of metabolites across known and emerging pathways bring functional clarity for some of these uncertain cases, connecting unexplained findings to meaningful diagnoses.



Global MAPS® Can Bring Your Patients Closer to Answers

Global MAPS® can be applied across a range of genetic testing outcomes to clarify findings and connect sequence data to functional evidence.

Global MAPS® adds functional biochemical insight that complements genomic findings by:

Clarifying uncertain or unexpected genetic findings, including variants of uncertain significance (VUS) or variants in metabolic genes without an obvious metabolic phenotype

Identifying metabolic disruptions when genetic testing alone does not explain the clinical presentation, helping uncover potential functional effects not captured by sequencing

Revealing novel or atypical metabolic pathway findings that may highlight additional gene-disease mechanisms

Transforming Ambiguous Results Into Actionable Insights

As part of Baylor Genetics Multimodal approach, Global MAPS® complements genome and exome sequencing by revealing how DNA changes may affect biochemical pathways to guide patient management.

● INCLUDED
○ AVAILABLE AS A SEPARATE TEST

	Whole Genome Sequencing	Whole Exome Sequencing
RNA-Seq Analysis	●	●
Complex Structural Variant (SV) Analysis	●	
Short Tandem Repeat (STR) Analysis	●	
Methylation Analysis of *FMR1*	●	
Mitochondrial DNA (mtDNA) Analysis	●	○
Copy Number Variants (CNV) Analysis	●	●
Uniparental Disomy (UPD) Analysis	●	
Metabolomic Analysis (Global MAPS®)*	○	○

* Metabolomic analysis (Global MAPS®) is available as a separate test for Whole Genome Sequencing and Whole Exome Sequencing and reported separately.

When to Order

Order Global MAPS® for patients with suspected metabolic or neurologic disease when:

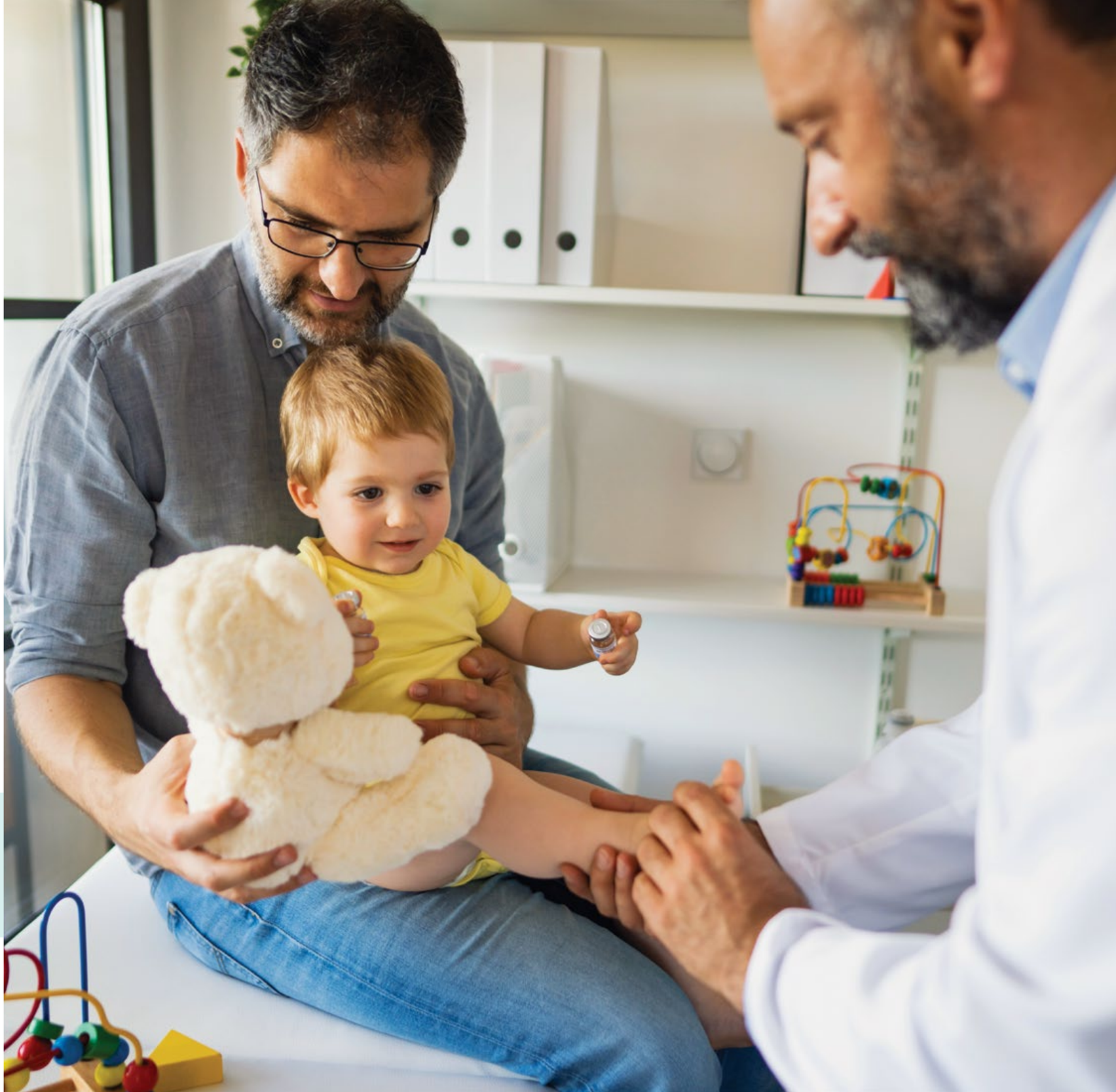
- Clinical features suggest an IEM, but conventional biochemical testing is inconclusive or uninformative
- Sequencing results are unclear or uncertain and require functional context
- A broad metabolic panel is needed early in the diagnostic journey due to overlapping, non-specific clinical presentation
- Novel, emerging, or atypical metabolic pathways need to be explored

The American Academy of Pediatrics recommends metabolic screening for global developmental delay and intellectual disability in children.[2]

Common Features of IEMs[4]

Developmental Delay or Regression
Intellectual Disability or Autism Spectrum Disorder
Abnormal Metabolic or MRI Findings
Unexplained Neurologic Symptoms or Seizures
Hypotonia
Hypoglycemia
Failure to Thrive
Recurrent or Persistent Vomiting

Every patient's diagnostic journey is unique. Global MAPS® may support providers at key decision points by bringing clarity when results are uncertain or incomplete.



Global MAPS® Test Features & Details

Results in 21 Days*

Test Name	Code	Sample
Global MAPS®–Plasma	4900	Frozen EDTA plasma
Global MAPS®–Urine	4901	Frozen Urine

Key Features

- Detects over 700 metabolites in a single test[5]
- Captures data across known and novel pathways
- Integrates into Baylor Genetics' multimodal approach (Whole Genome Sequencing/Whole Exome Sequencing, RNA Sequencing [RNA-Seq], and Metabolomics)

Data & Reporting

Global MAPS® provides semi-quantitative results using Z-scores that compare analyte levels against a healthy reference population. Rather than presenting isolated values, Global MAPS® reports highlight meaningful patterns of biochemical change to support clinical interpretation.

In addition to Z-score findings, Global MAPS® organizes results into four reporting categories to help providers understand the potential clinical relevance:

1. Significantly altered analytes: Metabolites with Z-scores outside the expected range that may relate to the patient's phenotype
2. Unusually **present** analytes: Rare metabolites detected in <5% of historical samples that may provide diagnostic clues
3. Unusually **absent** analytes: Metabolites typically present in >99% of samples but missing in this patient, suggesting potential pathway disruption
4. Analytes influenced by diet, supplements, or medications: Patterns that help distinguish true biochemical abnormalities from nutritional or treatment-related effects

Considerations and Limitations. Global MAPS® can detect metabolites that weigh from 75-1000 Da. Individual concentrations are not reported. Specific analytes will not automatically be reported; rather, analytes that altered, influenced, unusually present, or unusually absent are reported. Metabolomic testing cannot detect all genetic or biochemical conditions, including those with large molecule analytes. Interpretation depends on available knowledge of metabolic pathways and clinical context. Additional testing may be recommended for comprehensive evaluation. Results should always be interpreted in the context of the patient's clinical presentation and other laboratory findings. Global MAPS® is not intended for use in acute metabolic crises.

* Turnaround time can vary based on factors such as collection date, sample quality/quantity and completeness of patient information provided.

Conditions Assessed by Global MAPS®†

Adenylosuccinate Lyase Deficiency

AICA-Ribosiduria (ATIC Deficiency)

Argininemia

Argininosuccinic Aciduria

Aromatic L-Amino Acid Decarboxylase Deficiency

ß-Ureidopropionase Deficiency

Citrate Transporter Deficiency

Citrin Deficiency

Citrullinemia

Cobalamin Biosynthesis Disorders

Creatine Biosynthesis Defects (GAMT & AGAT)

DEGS1 Deficiency

Ethylmalonic Encephalopathy

GABA Transaminase Deficiency

Galactosemia

Glutaric Acidemia Type I

Glycerol Kinase Deficiency

Glycine Encephalopathies

Glycine N-Methyltransferase Deficiency

Holocarboxylase Synthetase Deficiency

Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Syndrome

Hyperphenylalaninemia

Isovaleric Acidemia

Kynurenine 3-Monooxygenase (KMO) Deficiency

Lysinuric Protein Intolerance

Maple Syrup Urine Disease

Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency

Methylmalonic Acidemia

Mitochondrial Neurogastrointestinal Encephalopathy (MNGIE)

MTHFR Deficiency

Ornithine Transcarbamylase Deficiency

Orotic Aciduria

Peroxisome Biogenesis Disorders / Zellweger Spectrum

Phenylketonuria

Primary Carnitine Deficiency

Propionic Acidemia

Pyridoxine-Dependent Epilepsy

Riboflavin Transporter Deficiency (SLC25A2)

Ribose-5-Phosphate Isomerase Deficiency

Serine Biosynthesis Disorders

Short Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency

Smith-Lemli-Opitz Syndrome

Spondyloepimetaphyseal Dysplasia, Genevieve Type

Succinic Semialdehyde Dehydrogenase (SSADH Deficiency

Thiamine Transporter Deficiency

Transaldolase Deficiency

Transketolase Deficiency

Trimethyllysine Hydroxylase Epsilon Deficiency

Tyrosinemia Type I

Urocanase Deficiency (Benign Condition)

Very Long Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency

Xanthurenic Aciduria (KYNU Deficiency)

2-Hydroxyglutaric Acidemia (Likely L-Form)

3-Hydroxy-3-Methylglutaryl (HMG)-CoA Lyase Deficiency

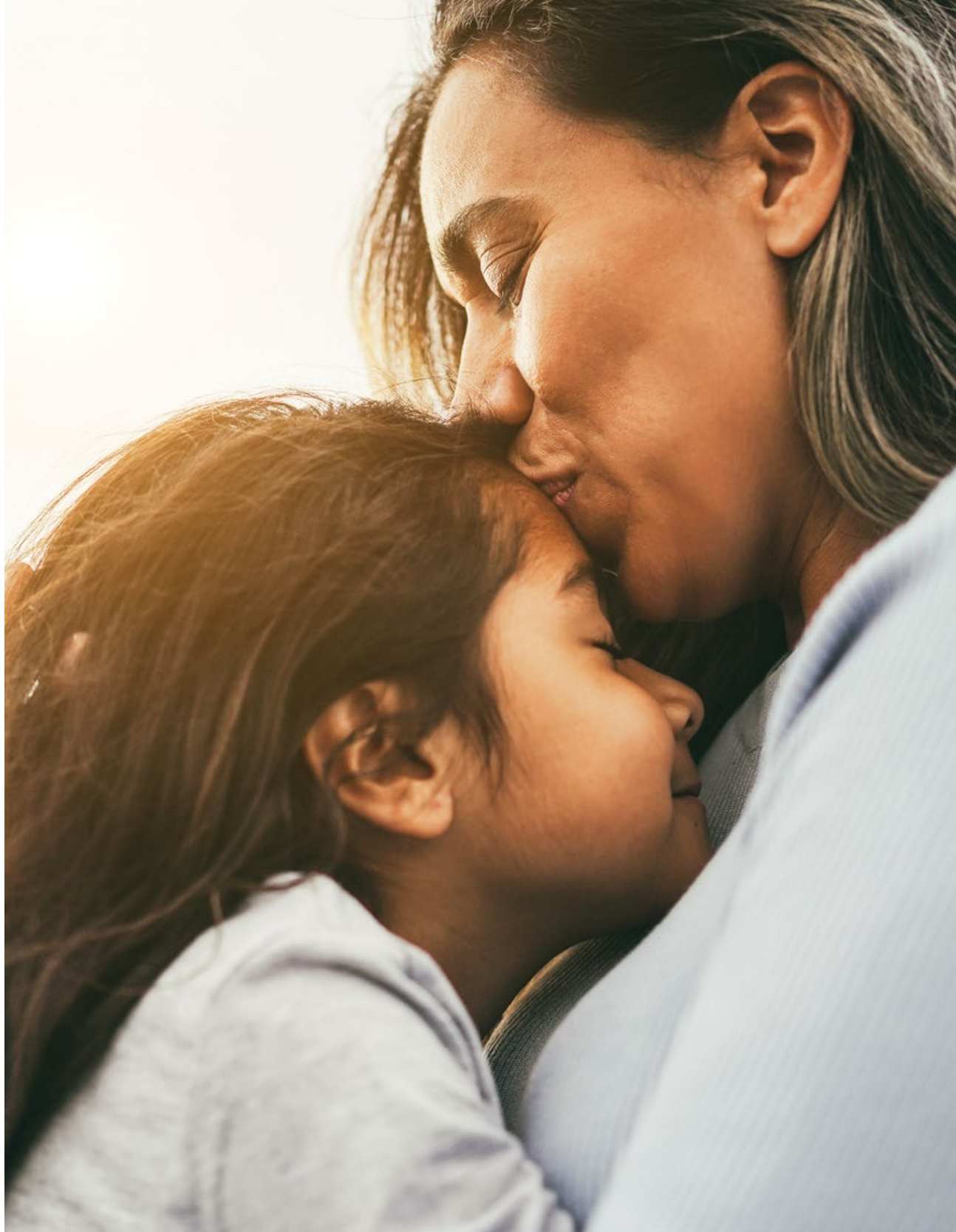
3-Hydroxyisobutyryl-CoA Hydrolase Deficiency (HIBCH)

3-Methylcrotonyl-CoA Carboxylase (3-MCC) Deficiency

† The conditions listed here represent those for which Global MAPS® has been validated and confirmed. Because Global MAPS® is a comprehensive metabolic-screening platform, it may also detect additional conditions or metabolic signatures beyond those listed, but these have not yet undergone the same level of validation or confirmation. Interpretation of screening results should always be performed in the context of clinical findings and, when appropriate, follow-up diagnostic testing.

References

1. PMID: 29083820
2. PMID: 40545261
3. PMID: 30600976
4. PMID: 37036266
5. Internal data
6. PMID: 7564553



45+ YEARS OF INNOVATION

4 MILLION+ CLINICAL TESTS PERFORMED

1 MILLION+ FAMILIES HELPED

1 MISSION EMPOWERING YOU WITH ANSWERS THAT MATTER

Baylor Genetics pioneered the history of genetic testing. Now, we're leading the way in precision diagnostics.

A pioneer of precision medicine for over 45 years, Baylor Genetics is a leading diagnostic genomics partner offering a full spectrum of clinically relevant genetic testing, including Whole Genome Sequencing, Whole Exome Sequencing, and focused panels. Baylor Genetics couples the fastest and most comprehensive precision diagnostics options with the support of genetic counselors to help clinicians and patients avoid a lengthy diagnostic odyssey, guide medical management, and make sure no patient with a genetic disorder gets left behind. Our test menu spans from family planning, pregnancy, neonatal and pediatric testing, oncology, and beyond.

Baylor Genetics is located in Houston's Texas Medical Center.

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