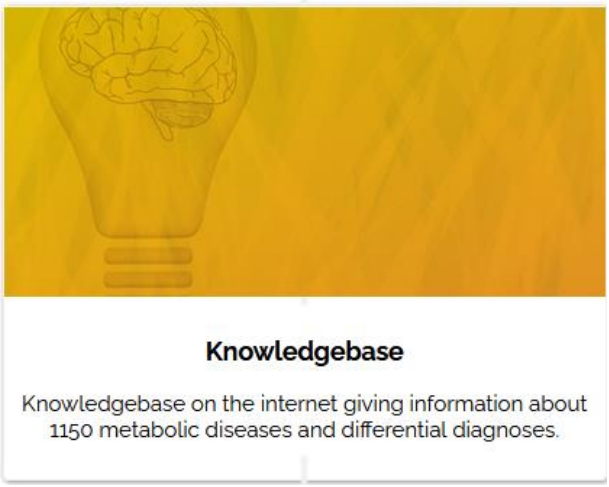
Metagene: KNOWLEDGEBASE



Metagene
Knowledgebase IEM

DISEASE SYMPTOM LAB

MIM	MIMGen	Disease	Synonym
202010	610613	11-BETA-HYDROXYLASE DEFICIENCY (CYP11B1)	ADRENAL HYPERPLASIA VI; STEROID 11 BETA-HYDROXYLASE DEFICIENCY
264300	605573	17-BETA HYDROXYSTEROID DEHYDROGENASE III DEFICIENCY	PSEUDOHERMAPHRODITISM, MALE, WITH GYNECOMASTIA; HSD17B3
616034	615787	2,4-DIENOYL-CoA REDUCTASE DEFICIENCY (DECRD)	2,4-DIENOYL-CoA REDUCTASE; DECRD; NADK2
204750	614984	2-AMINOADIPIC ACIDURIA (AMOCAD, AMOXAD)	AMINOADIPIC ACIDURIA; AMOXAD
600721	609186	2-HYDROXYGLUTARIC ACIDURIA (D) TYPE I	D-2-HYDROXYGLUTARIC ACIDURIA 1; D2HGA1
613657	147650	2-HYDROXYGLUTARIC ACIDURIA (D) TYPE II	D-2-HYDROXYGLUTARIC ACIDURIA 2; D2HGA2

Enter text in the search field DISEASE, e.g. pku for Phenylketonuria

DISEASE SYMPTOM LAB

MIM	MIMGen	Disease	Synonym
		pku	
261600	612349	PHENYLKETONURIA (PKU)	HYPERPHENYLALANINEMIA, MILD, INCLUDED; HPA, INCLUDED; PAH
261600		PHENYLKETONURIA, FETAL EFFECTS FROM MATERNAL (MPKU)	

Click PHENYLKETONURIA (PKU) to view details:

PHENYLKETONURIA (PKU)

Disease	PHENYLKETONURIA (PKU)
Synonym	HYPERPHENYLALANINEMIA, MILD, INCLUDED; HPA, INCLUDED; PAH
OMIM	261600 OMIM = Online Mendelian Inheritance of Men
Orphanet	79254
Protein (UniProt)	phenylalanine hydroxylase (PAH)
ExPASy	1.14.16.1
Gene locus	12q23.2 Detail information to gene locus by the National Center for Biotechnology Information NCBI: • Phenylalanine hydroxylase
TreatableID	https://treatable-id.net
ICD	E70.0, E70.1

Laboratory findings:

1. **2-Hydroxyphenylacetic acid increased (urine)**
2. **3-Phenyllactic acid increased (urine)**
3. **Phenylacetic acid increased (urine)**
4. **Phenylalanine increased (plasma)**
5. **Phenylalanine/Tyrosine increased (plasma)**
6. **Phenylpyruvic acid increased (urine)**
7. **Phenylalanine increased (urine)**
8. 4-Hydroxyphenyllactic acid increased (urine)
9. 5-Hydroxyindolacetic acid (5-HIAA) normal/dec (cerebrospinal fluid)
10. Biopterin increased (urine)
11. D-Mannitol normal/inc (urine)
12. Ferric chloride reaction positive (urine)
13. Homovanillic acid (HVA) normal/dec (cerebrospinal fluid)
14. L-Tyrosine decreased (urine)
15. N-Acetylphenylalanine increased (urine)
16. Neopterin increased (urine)
17. Phenylalanine increased (cerebrospinal fluid)

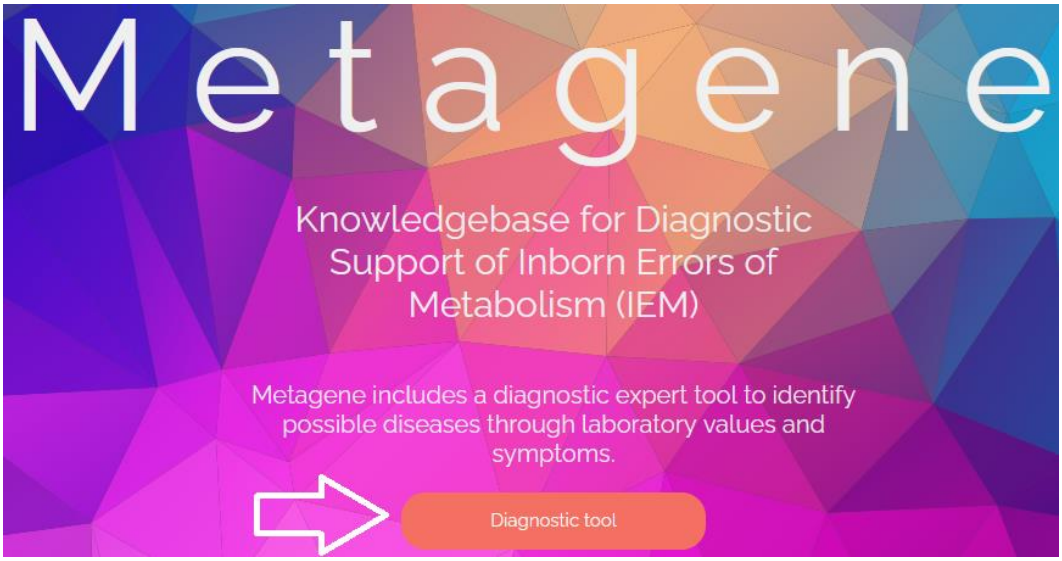
Symptoms:

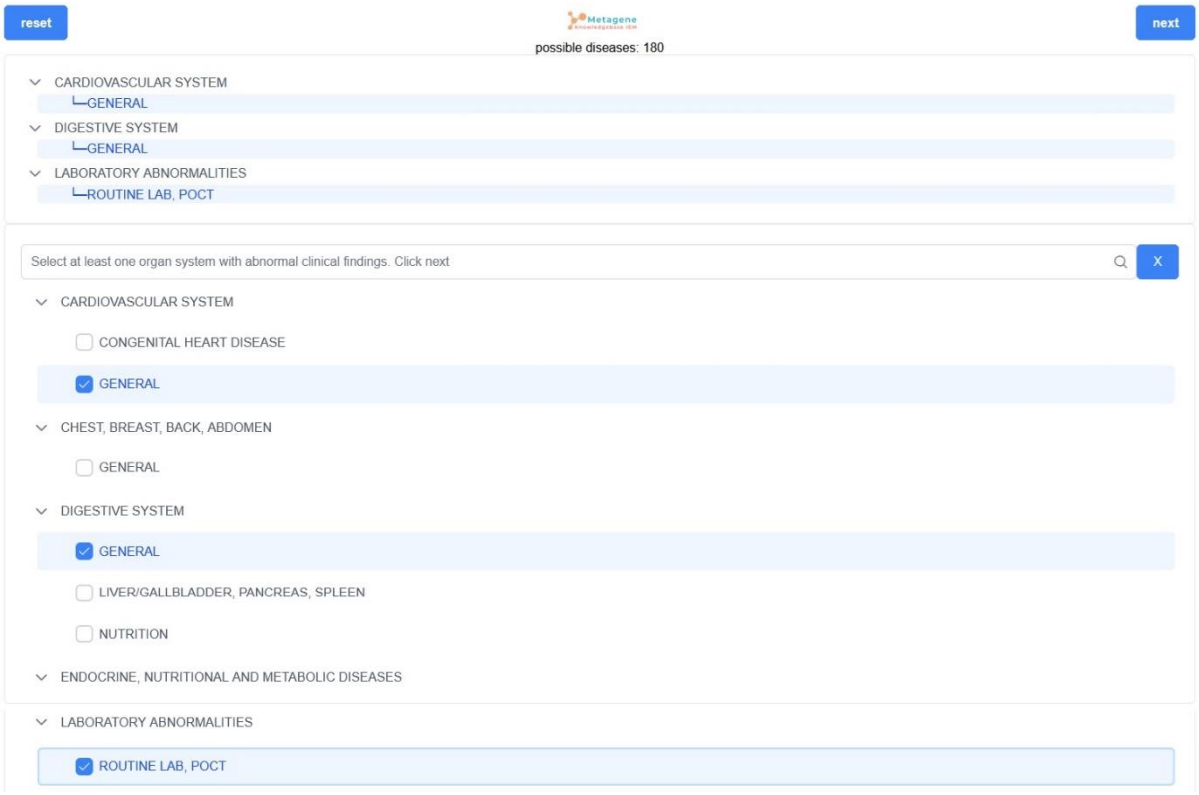
1. **intellectual disability/intellectual developmental disorder**
2. **musty body odor**
3. **blue eyes**
4. **fair hair**
5. **behavior, autism or autistic-like**
6. **decreased body height**
7. embryopathy
8. **hypertonia, spasticity**
9. hypopigmentation
10. **irritability**
11. **low birthweight (small for gestational age)**
12. **microcephaly (<2 SD for age)**
13. pigmentation, skin and sclera
14. seizures
15. **skin rash, eczematous or seborrhoic**
16. **unusual odor / odour**
17. **vomiting**
18. Amino acids, plasma
19. behavior, hyperactive, restless
20. behavior, self-mutilating or destructive
21. childhood
22. EEG abnormalities [-]
23. epilepsy
24. infancy
25. leukodystrophy
26. mental retardation
27. MRI, brain, white matter abnormalities [-]
28. newborn, neonatal
29. Organic acids, urine
30. sleep disturbances
31. small for gestational age (SGA), intrauterine growth retardation (IUGR)

Literature:

1. Autor: Susan E Waisbren, (2025)
   Beyond neuropsychological tests: AI speech analysis in PKU
2. Autor: François Feillet, (2025)
   Long-term safety of sapropterin in paediatric and adult individuals with phenylalanine hydroxylase deficiency: Final results of the Kuvan® Adult Maternal Paediatric European Registry multinational observational study
3. Autor: Ahring K, (2024)
   Management of phenylketonuria in European PKU centres remains heterogeneous
4. Autor: Feillet F, (2024)
   Efficacy and safety of sapropterin before and during pregnancy: Final analysis of the Kuvan® Adult Maternal Paediatric European Registry (KAMPER) maternal and Phenylketonuria Developmental Outcomes and Safety (PKUDOS) PKU-MOMs sub-registries
5. Autor: Trefz F, (2024)
   Does hyperphenylalaninemia induce brain glucose hypometabolism? Cerebral spinal fluid findings in treated adult phenylketonuric patients
6. Autor: Cary O Harding, (2024)
   Sepiapterin: a potential new therapy for phenylketonuria
7. Autor: Wendy E Smith, (2024)
   Phenylalanine hydroxylase deficiency diagnosis and management: A 2023 evidence-based clinical guideline of the American College of Medical Genetics and Genomics (ACMG)
8. Autor: WEBSITE MANAGER: Nenad Blau (2023)
   BioPKU
9. Autor: Alghamdi MA, (2023)
   Classical phenylketonuria presenting as maternal PKU syndrome in the offspring of an intellectually normal woman
10. Autor: Chen A, (2023)
    Clinical, genetic, and experimental research of hyperphenylalaninemia
11. Autor: Cannet C, (2023)
    Phenylketonuria (PKU) Urinary Metabolomic Phenotype Is Defined by Genotype and Metabolite Imbalance: Results in 51 Early Treated Patients Using Ex Vivo 1H-NMR Analysis
12. Autor: Muri R, (2022)
    Cortical thickness and its relationship to cognitive performance and metabolic control in adults with phenylketonuria
13. Autor: Dobrowolski SF, (2021)
    Phenylalanine hydroxylase deficient phenylketonuria comparative metabolomics identifies energy pathway disruption and oxidative stress
14. Autor: Dobrowolski SF, (2021)
    Mesenchymal stem cell energy deficit and oxidative stress contribute to osteopenia in the Pah enu2 classical PKU mouse
15. Autor: Evers RAF, (2020)
    Tetrahydrobiopterin treatment in phenylketonuria: A repurposing approach
16. Autor: Krämer J (2020)
    Case-control study about the acceptance of Pegvaliase in Phenylketonuria

Metagene: DIAGNOSE TOOL





Select at least one organ system or laboratory anomalies, e.g. CARDIOCASCULAR SYSTEM for arrhythmia, DIGESTIVE SYSTEM for vomiting, ROUTINE LAB for hypoglycemia. **Click “next”**

Search for and select clinical symptoms or laboratory anomalies one by one. **Click “next”.**

ICD	Disease	MIM
E71.1	3-HYDROXY-3-METHYLGLUTARYL-CoA LYASE DEFICIENCY (HMGCLD)	246450
E71.1	BARTH SYNDROME (BTHS)	302060
E71.3	CARNITINE PALMITOYL TRANSFERASE DEFICIENCY (II), CPT2, MYOPATHIC, STRESS-INDUCED	255110
E71.3	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY (CACT)	212138
H49.8	KEARNS-SAYRE SYNDROME (KSS)	530000
E71.3	LONG-CHAIN-3-HYDROXYACYL-CoA DEHYDROGENASE DEFICIENCY (LCHAD)	609016
E71.3	MEDIUM CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY (MCAD)	201450
E71.1	PROPIONIC ACIDEMIA (PA, PCCA)	606054
E71.1	PROPIONIC ACIDEMIA (PA, PCCB)	606054

9 diseases with cardiac arrhythmia, vomiting and hypoglycemia.