ProBabyDNA

Baby Sensor

Musterfrau Manuela

DEMO_ML

ProGenom
www.progenom.com

COVER LETTER

Dear Ms. Musterfrau,

Your sample for the analysis arrived on 25/11/2020 in the laboratory and was evaluated according to the highest laboratory quality standards. The results were evaluated and released by two independent geneticists and molecular biologists. After obtaining the results, your personal report was compiled. We hereby convey the results to you in the format of your choice.

We would like to thank you for your trust and hope that you are satisfied with our service. We are always open to questions and suggestions. Please do not hesitate to contact us. We value your feedback. This is the only way we can continuously improve our services.

We hope the analysis meets your expectations.

Kind regards,

Dr. Daniel Wallerstorfer BSc.
Laboratory Director

Florian Schneebauer, MSc.
Laboratory Manager

Baby Sensor

Analysis report
Musterfrau Manuela | Date of birth: 01/01/2015

Order number:
DEMO_ML

This report contains personal medical information that is highly confidential. Data protection must be ensured.

SCOPE OF ANALYSIS

Here you can find out which diseases were analyzed by our test

Scope of the Analysis

This test analysed the urine sample of your child for more than 100 genetic and metabolic disorders and is one of the most comprehensive tests of its kind.

Here is a list of diseases covered by Baby Sensor, which can either be prevented or treated if diagnosed in time.

Disease	Complications	Type
Transient Galactosemia	poor weight gain coincident with liver dysfunction	Type A
2-ketoadipic Aciduria	Developmental delay and other neurological problems	Type A
3-hydroxy-3-methylglutaric aciduria (HMG CoA lyase deficiency)	severe metabolic acidosis without ketosis / Developmental delay	Type A
3-hydroxyisobutyryl CoA decarboxylase deficiency	severe hypoketotic hypoglycemia and neurological damage	Type B
3-methylcrotonyl CoA carboxylase (3-MCC) deficiency	Acute metabolic acidosis, delayed development	Type A
3-methylglutaconic Aciduria	Cardiomyopathy and skeletal myopathy	Type B
5-oxoprolinuria	seizures, mental retardation and a loss of coordination	Type A
Hyperglycinuria(ketotic)	Failure to thrive	Type A
Adenine phosphorybosyl transferase deficiency	Urinary tract infection	Type B
Adenosine deaminase deficiency	Episodes of (otitis) ear infections and upper respiratory tract infections	Type B
Alkaptonuria	Damage to cartilage	Type A
Argininemia	Loss of developmental milestones	Type A
Argininosuccinic aciduria	Mental and motor retardation	Type A
Biotinidase deficiency	Scaly perioral/facial rash / mental retardation	Type A
Canavan disease	Severe regression of milestones	Type B
Hyperuric acidemia	Sensorineural hearing impairment, Uric acid urolithiasis	Type A
Citrullinemia	Lethargy and abnormal behavior	Type A
Cystathionuria	Liver dysfunction	Type A
Cystinuria	Delay in development	Type A
Benign hyperphenylalaninemia	eczema and fair hair and skin coloring	Type A
Methylmalonic aciduria, cblA and cblB forms (MMA, Cbl, A,B)	weak muscle tone (hypotonia), developmental delay, excessive tiredness (lethargy), an enlarged liver	Type A

Disease	Complications	Type
D-glyceric aciduria	Poor weight gain	Type C
Endogenous sucroseuria	Mental retardation	Type B
Dihydropyridinase Deficiency	Neonatal convulsions	Type D
Dihydrolipoyl dehydrogenase (E3) deficiency	Sweet smell to urine and body - like burnt sugar	Type D
Ethyl malonic aciduria	Failure to thrive / coma	Type D
Formiminoglutamic aciduria	Megaloblastic anemia	Type A
Fructose-1,6-diphosphatase deficiency	Hypoglycemia with ketosis	Type A
Fumarate hydratase deficiency	Seizures with severe retardation	Type D
Galactosemia	Hepatic dysfunction	Type A
Glutaric aciduria type I	Large head with movement difficulty	Type A
Glutaric aciduria type II	Sweaty feet odor and breathing problems / Mental and motor retardation	Type B
Glutathionuria	Hemolytic anemia	Type B
Beta- ketothiolase deficiency (BKT)	vomiting, dehydration, difficulty breathing, extreme tiredness (lethargy) and occasionally seizures	Type A
Primary hyperoxaluria type 2	Kidney stones, kidney damage, kidney failure, and injury to other organs	Type A
Hartnup Disease	Sensitivity to light and eye defect	Type A
Hawkinsonuria	Hepatic dysfunction	Type C
Histidinemia	Mental retardation, Renal defect	Type C
Histidinuria	Mental retardation different facial features	Type A
Homocystinuria	Mental retardation	Type A
Hydroxylysinuria	Mental retardation, behavioral problems and hyperactivity	Type C
Hyper hydroxyprolinemia	Mental retardation	Type C
Hyperglycinuria	Failure to thrive	Type B
Hyperleucine-Isoleucinemia	Seizures, failure to thrive	Type B
Hypermethioninemia	Unusual facial features, neurological problems, motor developmental delay	Type A
Hyperornitininemia Hyperammonemia-hyperhomocitrullinemia (HHH) syndrome	Vomiting,lethargy, developmental delay, learning disabilities	Type D
Hyperpipecolatemia	Severe delayed development	Type C
Hyperprolinemia Type-1	Neurological or psychiatric problems	Type C
Hyperprolinemia Type-II	Seizures, mental retardation	Type C
Imidiazole amino aciduria	Seizures, delayed development	Type B
Iminoglycinuria	Mental retardation and kidney problems	Type C
Infantile refsum disease	Blindness and hearing problems / retinitis pigmentosa	Type C
Isovaleric Acidemia	Twitching due to hypothermia, ‘sweaty feet’ odor	Type A
Lactose intolerance	Delayed development	Type A
Leigh syndrome	General weakness with heart problems	Type C
Lesch-Nyhan syndrome	Mental retardation / self biting habit	Type C
Long chain acyl-CoA dehydrogenase deficiency	Muscle weakness / Consistent muscle pain	Type A

Disease	Complications	Type
Lysinuria	Mental retardation	Type C
Lysinuric Protein Intolerance	Poor weight gain	Type B
Maple Syrup Urine Disease	Neurological impairment, lethargy, Sweet smell to urine and body - like burnt sugar	Type A
Medium chain acyl CoA dehydrogenase deficiency	Failure to thrive	Type A
Methylmalonic Acidemia	Convulsions, lethargy	Type A
Methylmalonic Acidemia (MMA) due to abnormal metabloism, absorbtion & transport of Vitamin B12	Muscle weakness, convulsions and anemia	Type A
Multiple Carboxylase Deficiency (MCD)	Metabolic acidosis, decreased muscle tone, developmental delay	Type A
Mevalonic acidemia	Abnormal head shape, delayed developmental milestones	Type B
Methylmalonic semialdehyde dehydrogenase (MMSDH) deficiency	Metabolic acidosis, lethargy, seizures	Type B
N-acetylglutamate synthetase (NAGS) deficiency	Neurologic complications	Type B
Neonatal Adrenoleucodystrophy	Weakening of muscles	Type D
Neuroblastoma	Spontaneous regression	Type A
NICCD	Hepatic dysfunction	Type A
Ornithine transcarbamylase (OTC) deficiency	Irritational behaviour developmental delay	Type A
Orotic aciduria	Heart malformation and anemia	Type B
Partial deficiency of hypoxanthine-adenine phosphorybosyl transferase deficiency	Kidney stones / Movement problems	Type D
Phenylketonuria (PKU)	Developmental delay and behaviour problems	Type A
Primary hyperoxaluria	Renal colic with urinary stones	Type A
Propionic acidemia	Lethargy, poor feeding, hypotonia	Type A
Glycerol Kinase Deficiency	Growth Retardation	Type A
Pyruvate carboxylase deficiency	Respiratory problems,	Type D
Pyruvate decarboxylase deficiency	developemental disorder, severe movement disorder and vision problems	Type D
Pyruvate dehydrogenase (E1) deficiency	Poor feeding, lethargy and respiratory problems	Type D
Pyruvate dehydrogenase phosphatase deficiency	Lactic acidosis with decreased muscle tone	Type D
Saccharropinuria	Dysmorphic features, Short stature	Type C
carbamoyl phosphate synthetase 1-deficiency	Neurologic complications	Type B
Tyrosinemia Type III	Mild mental retardation, Convulsions, balancing difficulty	Type B
Serum carnosinase deficiency	Decreased muscle tone delayed development	Type C
Short chain acyl CoA dehydrogenase deficiency	Low blood sugar, lethargy	Type A
Citrullinemia type II	restlessness, memory loss, abnormal behaviors, seizures, and coma	Type B
Thymine Uraciluria	Mental retardation	Type D
Transient tyrosinemia in infancy	Prolonged jaudice, lethargy	Type A

Disease	Complications	Type
Tryptophanuria with dwarfism	Short stature, mental retardation	Type B
Tyrosinemia due to liver dysfunction	Hepatic dysfunction	Type A
Tyrosinemia Type I	Cabbage-like odor to urine, liver dysfunction	Type A
Tyrosinemia Type II	Eyes sensitive to light, delayed development	Type B
Valinemia	Failure to thrive, vomiting and developmental delay	Type D
Xanthinuria	Acute renal failure	Type A
Xanthurenic aciduria	Mental retardation	Type A
Zellweger like syndrome	Decreased muscle tone, severe psycho-motor retardation	Type D
Zellweger syndrome	Decreased muscle tone, dysmorphic features	Type D
β-aminoisobutyric aciduria	Neurological impairment	Type D
Defects of biopterin cofactor biosynthesis (BIOPT BS)	Developmental delays,Seizures (known as epilepsy), Behavioral troubles	Type B
Defects of biopterin cofactor regeneration (BIOPT REG)	intellectual disability,movement disorders, difficulty swallowing, seizures	Type B
Galactokinase deficiency (GALK)	development of cataracts,enlarged liver and spleen	Type B
Galactose epimerase deficiency (GALE)	delayed growth and development, intellectual disability, liver disease, and kidney problem	Type B
Isobutyryl-CoA dehydrogenase deficiency (IBD)	anemia, weak muscle tone, developmental delay	Type B
Malonic acidemia (MAL)	Hypoglycemia, vomiting, diarrhea, seizures	Type B
Methylmalonyl- CoA mutase deficiency (MUT)	severe keto acidosis, psychomotor dysfunction, dystonia	Type B
Mitochondrial trifunctional protein Deficiency	Lethargy, hypoglycemia, weak muscle tone (hypotonia), liver problems	Type B
Iminoglycinuria	Mental retardation and kidney problems	Type C
Fructosuria	Hepatomegaly, jaundice, cirrhosis, convulsions, failure to thrive and mental retardation	Type C
Hypersarcosinemia	visual impairment, cardiomyopathy, cranial synostosis, growth and mental retardation	Type C
Succinic semialdehyde dehydrogenase deficiency	Weak muscle tone (hypotonia), weak reflexes, seizures and a nonprogressive gait disturbances	Type D
Histidinuria	Mental retardation, different facial features	Type D
Aminoacylase I Deficiency	Deafness, Muscle weakness, Cerebral Atrophy	Type D

Type A Prevention and treatment measures are very effective for this disorder. In most cases the baby can be completely healthy or suffer only from minor complications of diagnosed early and treated in time.

Type B Prevention for these disorders is more difficult, but right diagnosis helps in prevention or treating acute complications for the best possible outcome for the child. Harmful and potentially fatal episodes of the disorder can often be prevented or treated effectively to improve complications.

Type C These disorders cause compliactions in only about 10% of affected individuals, which need to be treated. 90% of individuals exhibit no symptoms at all.

Type D These disorders are more severe and may significantly impact the babies health even with early diagnosis. Correct diagnosis answers many urgent questions when complications arise and enable supportive therapy to reduce symptoms and complications.

RESULT

Find out whether a genetic disease was detected here

The result

You had the urine sample of your child tested for signs for 101 genetic disorders and the results have been successfully analysed by our geneticists and molecular biologists.

No disorder identified

We have measured all relevant parameters and the analysis revealed no signs for one of the 101 genetic disorders covered by this test.

As a consequence, your child does not require special preventive measures or treatment besides a generally healthy lifestyle.

Please note:

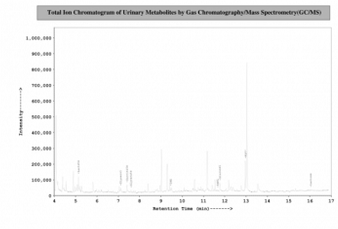
This test only covers the genetic disorders in the portfolio of Baby Sensor 100+. Other disorders cannot be detected by this method and so this report can only give information on the status of any of the 101 diseases. As is the case in any laboratory procedure, the detection rate is approximately 99% and the results apply only to the urine sample you provided for analysis. Diseases, that develop later in life are not covered by this test, but are available separately for you and your children.

Technical details

In the previous sections you have seen the technical result interpretation done by our metabolic experts and scientists. In this section you see the actual raw data of the analysis. This will enable external metabolic experts to review the analysis results and confirm the diagnosis.

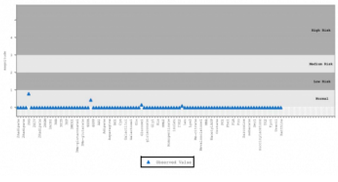
What do the graphs mean?

Technical reports, especially those of an advanced thechnology such as GC/MS are difficult to interpret and it takes years of study to fully understand and interpret them correctly. Here is just a short explanation of the elements of the results.



The Chromatogram

This image shows all of the substances present in your Baby's urine. The different substances are shown next to each other (along the horizontal axis) and the high peaks show how much of each of these substances is present in the urine sample.



Values close to or in the risk area

During this analysis we have measured 250 different substances in your Baby's urine. From scientific studies we know which substances are too high in a babies urine when one of the 101 metabolic diseases is present. With the technology we are using, this is quite easy to determine, as it is usually 5 to 8 substances that are too high at the same time. This allows fast and easy diagnosis of the right disorder in one go. If one or a few substances are in the risk area, this is quite common and no reason to worry. Only when the right substances are too high in combination, this means there is a disease present.

Sr. No	Metabolite Name	Control Value	Observed Value
1	2Aadipate	0.637	N.D
2	2E3HP	0.547	N.D
3	2Hadipate	0.619	N.D
4	2HB	0.100	N.D
5	2HG	5.074	4.013
6	2HIC	0.100	N.D
7	2HIV	0.760	N.D
8	2K3MV	0.100	N.D

"Observed Value" and "Control Value"

The "Observed Value" is the amount that was measured in your Baby's urine. In many cases the result will say "N.D", which means that the substance was present in such low amounts, that the machine was unable to detect any. This is a good sign. The "Control Value" is a standard (a certain threshold) that our scientists use to compare the results of your Baby. If the observed value is close to or higher than these standard values, our scientists pay particular attention to these substances when interpreting the results.

Name	[redacted]	**Age/ Gender**	[redacted]
Sample	Urine Sample	**Report Date**	[redacted]

Screening for 111 metabolic disorders by Urinary Total Ion Chromatogram Analytics (UTICA)

Result

Sample is screened negative for disorders tested. Please correlate the results with other clinical and diagnostic findings.

Note

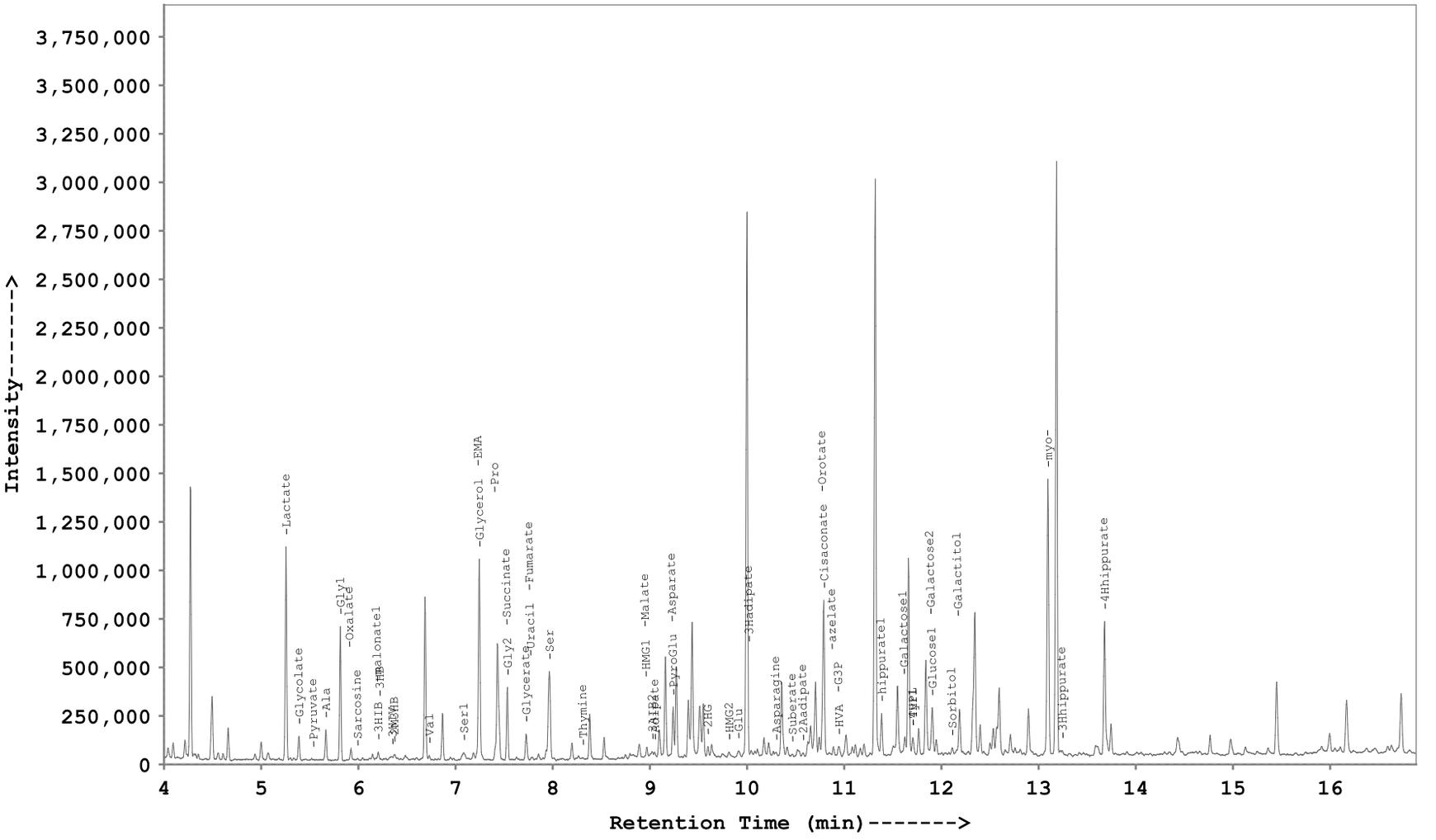
** This result is with reference to GC/MS analysis (List Enclosed) of the urine sample received this time only and does not rule out all IEMs (other than those screened for)*

** A possibility of 2% human error is possible*

** The clinician is requested to correlate this report with other clinical, radiological and laboratory findings*

** Some metabolites may be found to be increased in several conditions other than IEMs such as in case of administered drugs / IV fluids etc. So, it is important to take into consideration such conditions during interpreting the report*

Total Ion Chromatogram of Urinary Metabolites by Gas Chromatography/Mass Spectrometry(GC/MS)



Annexure

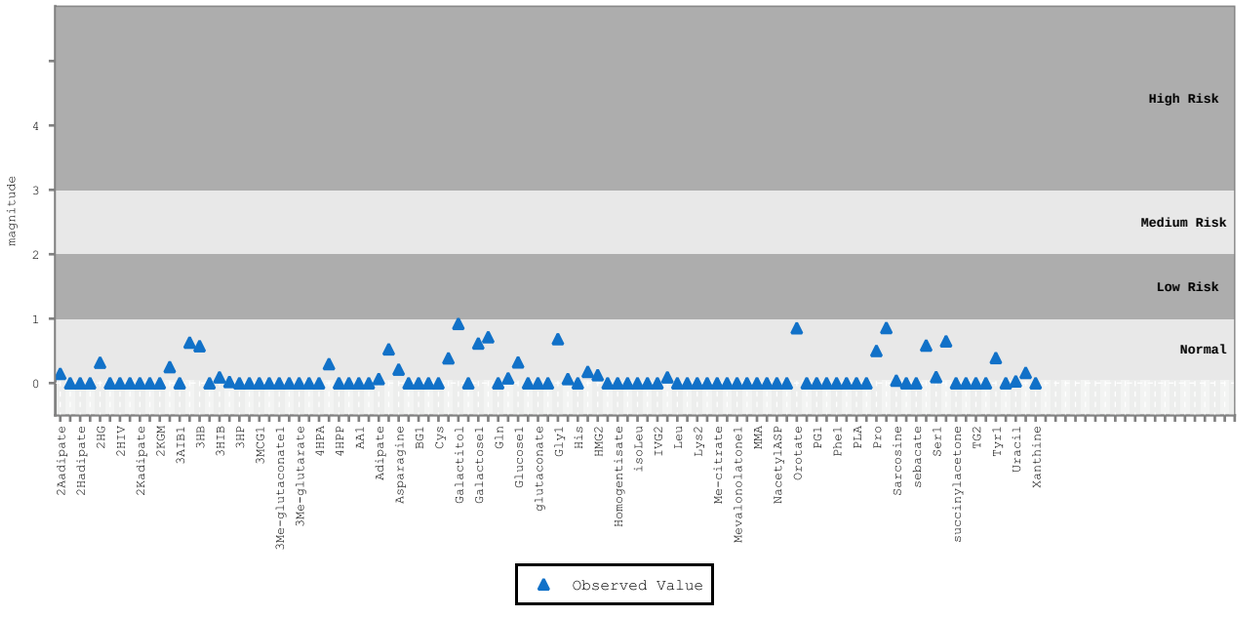
Detailed report of the tested disorders under Amino Acidopathies, Organic Acidemias, TCA Cycle , Mitochondrial Abnormality, Fatty Acid Metabolism, Peroxisomal, Purine & Pyrimidine metabolism, Sugar metabolism and Non-IEM disorders are illustrated in the table below. The graph of metabolic biomarkers used is shown below. Metabolic Biomarkers associated with the disorder are given with their control and observed values.

1. Amino Acidopathies and Organic Acidemias:

Sr. No	Name of the Disorder	Result
1	Beta- ketothiolase deficiency (BKT)	Negative
2	Alkaptonuria	Negative
3	Argininemia	Negative
4	Biotinidase deficiency	Negative
5	carbamoyl phosphate synthetase 1-deficiency	Negative
6	Dihydrolipoyl dehydrogenase (E3) deficiency	Negative
7	Familial Renal iminoglycinuria	Negative
8	Glutaric aciduria type I	Negative
9	Glutaric aciduria type II	Negative
10	Hartnup Disease	Negative
11	Homocystinuria	Negative
12	Hyperhydroxyprolinemia	Negative
13	3-hydroxy-3-methylglutaryl-CoA-lyase deficiency	Negative
14	Hyperleucine-isoleucinemia	Negative
15	Iminoglycinuria	Negative
16	Isovaleric acidemia	Negative
17	2-ketoadipic aciduria	Negative
18	Lysinuric protein intolerance	Negative
19	Maple syrup urine disease (MSUD)	Negative
20	3-methylcrotonyl CoA carboxylase deficiency	Negative
21	3-methylglutaconic aciduria	Negative
22	Methylmalonic semialdehyde dehydrogenase deficiency	Negative
23	Mevalonic aciduria	Negative
24	Ornithine transcarbamylase deficiency	Negative
25	Phenylketonuria (PKU)	Negative
26	Propionic acidemia	Negative
27	Transient neonatal tyrosinemia	Negative
28	Tyrosinemia Type I	Negative

Sr. No	Name of the Disorder	Result
29	Valinemia	Negative
30	Canavan disease	Negative
31	Multiple carboxylase deficiency	Negative
32	Hyperglycinuria(non-ketotic)	Negative
33	Hypersarcosinemia	Negative
34	Hypermethioninemia	Negative
35	Argininosuccinic aciduria	Negative
36	Citrullinemia Type 1	Negative
37	Cystathioninuria	Negative
38	Hyperornithinemia-hyperammoninemia-hyperhomocitrullinemia (HHH) syndrome	Negative
39	Hyperprolinemia type I	Negative
40	Hyperprolinemia type II	Negative
41	Saccharopinuria	Negative
42	Tyrosinemia Type II	Negative
43	Tyrosinemia Type III	Negative
44	Xanthurenic aciduria	Negative
45	Formiminoglutamic aciduria	Negative
46	Glutathionuria	Negative
47	Histidinemia	Negative
48	Serum carnosinase deficiency	Negative
49	Histidinuria	Negative
50	Hydroxylysinuria	Negative
51	Tryptophanuria with dwarfism	Negative
52	beta-aminoisobutyric aciduria	Negative
53	Hyperpipecolatemia	Negative
54	Imidazole amino aciduria	Negative
55	Hyperglycinuria(ketotic)	Negative
56	3-hydroxyisobutyryl CoA deacylase deficiency	Negative
57	Defects of biopterin cofactor biosynthesis (BIOPT BS)	Negative

58	Defects of biopterin cofactor regeneration (BIOPT REG)	Negative
59	NICCD	Negative
60	Benign hyperphenylalaninemia	Negative
61	Cystinuria	Negative
62	Citrullinemia type II	Negative
63	Methylmalonic acidemia (MMA) - Cbl C, D	Negative
64	Malonic acidemia (MAL)	Negative
65	Methylmalonic aciduria, cblA and cblB forms (MMA, Cbl A,B)	Negative
66	Methylmalonyl- CoA mutase deficiency (MUT)	Negative
67	Isobutyryl-CoA dehydrogenase deficiency (IBD)	Negative
68	Aminoacylase I Deficiency	Negative
69	Succinic semialdehyde dehydrogenase deficiency	Negative
70	Hawkinsunuria	Negative
71	Lysinuria	Negative
72	N-acetylglutamate / Carbamylphosphate synthetase deficiency	Negative
73	5-oxoprolinuria	Negative
74	Tyrosinemia caused by liver dysfunction	Negative



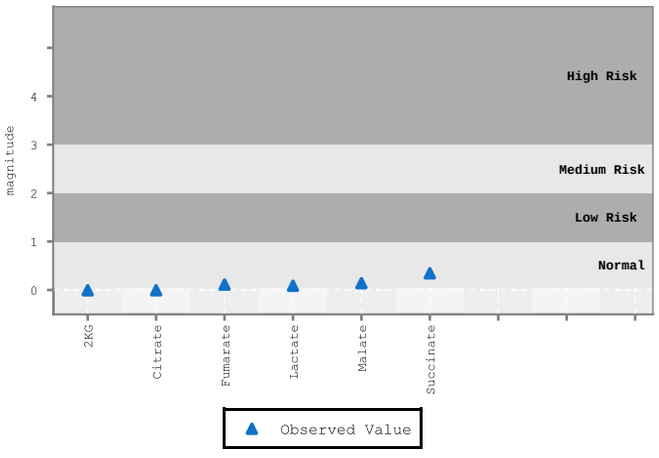
Sr. No.	Metabolite Name	Control Value	Observed Value
1	2Aadipate	0-5	0.727
2	2E3HP	0-0.7	N.D
3	2Hadipate	0-1.2	N.D
4	2HB	0-0.1	N.D
5	2HG	0.2-7.1	2.257
6	2HIC	0-0.1	N.D
7	2HIV	0-0.1	N.D

Sr. No.	Metabolite Name	Control Value	Observed Value
8	2K3MV	0-0.1	N.D
9	2Kadipate	0-0.1	N.D
10	2KG	0-1.1	N.D
11	2KGM	0-6.4	N.D
12	2M3HB	0-3	0.755
13	3AIB1	0-39	N.D
14	3AIB2	0-0.5	0.301

15	3HB	0-0.1	0.058	50	Glutarate	0-0.4	N.D
16	3HG	0-0.1	N.D	51	Gly1	0.1-28.9	19.793
17	3HIB	0-3.3	0.308	52	Gly2	0-25.7	1.692
18	3HIV	0-10.9	0.222	53	His	0-27.6	N.D
19	3HP	0-1.1	N.D	54	HMG1	0-0.8	0.147
20	3Hsebacate	0-1.8	N.D	55	HMG2	0.1-2.2	0.274
21	3MCG1	0-0.1	N.D	56	Homo-Cys	0-0.1	N.D
22	3MCG2	0-0.1	N.D	57	Homogentisate	0-0.1	N.D
23	3Me-glutaconate1	0-0.2	N.D	58	HomoSer	0-13.6	N.D
24	3Me-glutaconate2	0-0.2	N.D	59	isoLeu	0-5.6	N.D
25	3Me-glutarate	0-0.1	N.D	60	IVG1	0-0.1	N.D
26	4HB	0-0.1	N.D	61	IVG2	0-0.1	N.D
27	4HPA	0.1-10	N.D	62	Lactate	1.1-208.1	19.121
28	4HPL	0-3.7	1.092	63	Leu	0-9.3	N.D
29	4HPP	0-0.1	N.D	64	Lys1	0-8.8	N.D
30	4Hpro	0-18	N.D	65	Lys2	0-5.5	N.D
31	AA1	0-0.1	N.D	66	Mandelate	0-0.1	N.D
32	AA2	0-0.1	N.D	67	Me-citrate	0-2.4	N.D
33	Adipate	0.1-7.2	0.474	68	Met	0-1	N.D
34	Ala	0.4-25	13.201	69	Mevalonolatone1	0-0.1	N.D
35	Asparagine	0-5.3	1.133	70	Mevalonolatone2	0-0.1	N.D
36	b-Ala	0-0.2	N.D	71	MMA	0-0.4	N.D
37	BG1	0-0.1	N.D	72	N-AcetyTyr	0-0.1	N.D
38	BG2	0-0.1	N.D	73	NacetylASP	0-8.3	N.D
39	Cys	0-2	N.D	74	Orn	0-0.9	N.D
40	Dimethylglycine	0.1-1.7	0.678	75	Orotate	0-0.1	0.086
41	Galactitol	0-7.7	7.092	76	PA	0-0.3	N.D
42	Galactonate	0-10.6	N.D	77	PG1	0-0.2	N.D
43	Galactose1	0-0.4	0.25	78	PG2	0-0.1	N.D
44	Galactose2	0-1.4	1.031	79	Phe1	0-0.1	N.D
45	Gln	0-1.1	N.D	80	Phe2	0-8.5	N.D
46	Glu	0-7.4	0.565	81	PLA	0-0.1	N.D
47	Glucose1	0-35.3	11.459	82	PPA	0-0.1	N.D
48	Glucose2	0.4-27.5	N.D	83	Pro	0-19.4	9.728
49	glutaconate	0-0.1	N.D				

2. TCA Cycle/Mitochondrial Abnormality:

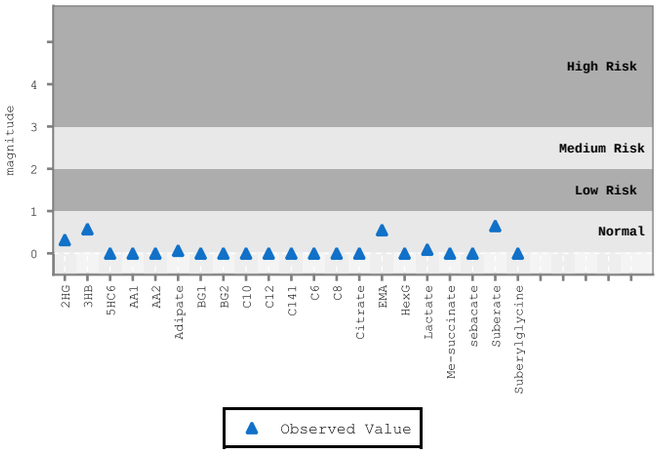
Sr. No	Name of the Disorder	Result
1	Fumarate hydratase deficiency	Negative
2	Pyruvate decarboxylase deficiency	Negative
3	Pyruvate carboxylase deficiency	Negative
4	Pyruvate dehydrogenase (E1) deficiency	Negative
5	Pyruvate dehydrogenase phosphatase deficiency	Negative
6	Leigh syndrome	Negative



Sr. No.	Metabolite Name	Control Value	Observed Value
1	2KG	0-1.1	N.D
2	Citrate	0-10.3	N.D
3	Fumarate	0-4	0.47
4	Lactate	1.1-208.1	19.121
5	Malate	0-2.9	0.422
6	Succinate	0-4.7	1.631

3. Disorders of Fatty Acid Metabolism:

Sr. No	Name of the Disorder	Result
1	Medium chain acyl CoA dehydrogenase deficiency	Negative
2	Short chain acyl CoA dehydrogenase deficiency	Negative
3	Ethyl Malonic Aciduria	Negative
4	Mitochondrial trifunctional protein Deficiency	Negative
5	Glycerol Kinase Deficiency	Negative
6	Very Long-chain acyl-CoA dehydrogenase deficiency	Negative

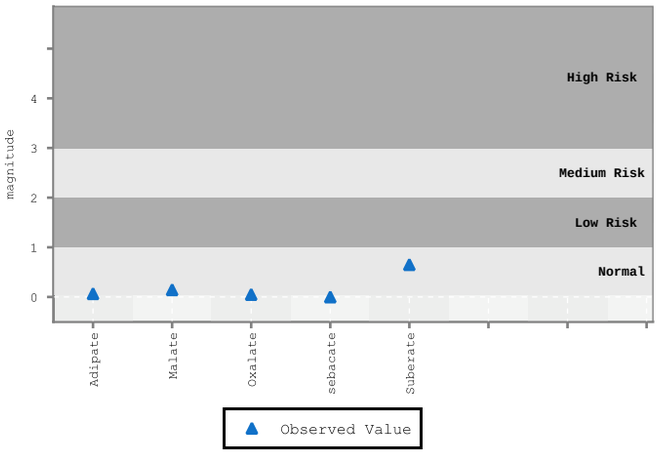


Sr. No.	Metabolite Name	Control Value	Observed Value
1	2HG	0.2-7.1	2.257
2	3HB	0-0.1	0.058
3	5HC6	0-0.8	N.D
4	AA1	0-0.1	N.D
5	AA2	0-0.1	N.D
6	Adipate	0.1-7.2	0.474
7	BG1	0-0.1	N.D
8	BG2	0-0.1	N.D
9	C10	0-0.1	N.D
10	C12	0-0.1	N.D

Sr. No.	Metabolite Name	Control Value	Observed Value
11	C141	0-0.1	N.D
12	C6	0-5.9	N.D
13	C8	0-0.1	N.D
14	Citrate	0-10.3	N.D
15	EMA	0-0.1	0.055
16	HexG	0-0.5	N.D
17	Lactate	1.1-208.1	19.121
18	Me-succinate	0-0.1	N.D
19	sebacate	0-0.1	N.D

4. Peroxisomal Disorders:

Sr. No	Name of the Disorder	Result
1	Zellweger like syndrome	Negative
2	Zellweger syndrome	Negative
3	Primary hyperoxaluria type 2	Negative
4	Infantile refsum disease	Negative
5	Neonatal Adrenoleukodystrophy	Negative
6	Primary hyperoxaluria Type 1	Negative

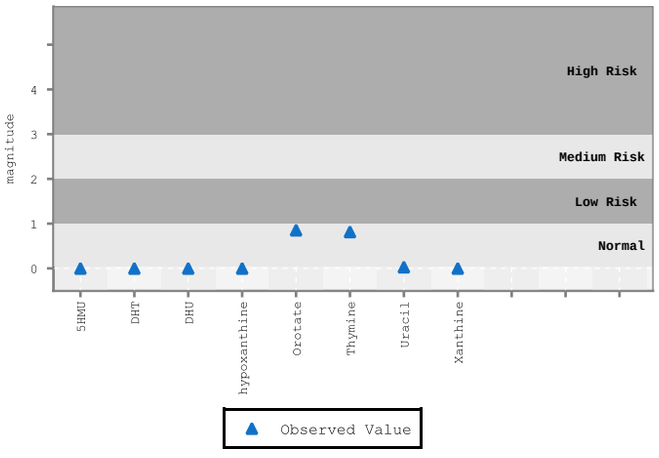


Sr. No.	Metabolite Name	Control Value	Observed Value
1	Adipate	0.1-7.2	0.474
2	Malate	0-2.9	0.422
3	Oxalate	0-2.8	0.141

Sr. No.	Metabolite Name	Control Value	Observed Value
4	sebacate	0-0.1	N.D
5	Suberate	0-0.5	0.327

5. Disorders of Purine and Pyrimidine Metabolism:

Sr. No	Name of the Disorder	Result
1	Lesch-Nyhan syndrome	Negative
2	Dihydropyrimidinase Deficiency	Negative
3	Orotic aciduria	Negative
4	Thymine Uraciluria	Negative
5	Xanthinuria	Negative
6	Adenosine deaminase deficiency	Negative
7	Adenine phosphoribosyl transferase deficiency	Negative
8	Partial deficiency of hypoxanthine-guanine phosphoribosyltransferase deficiency	Negative
9	Hyperuric acidemia	Negative

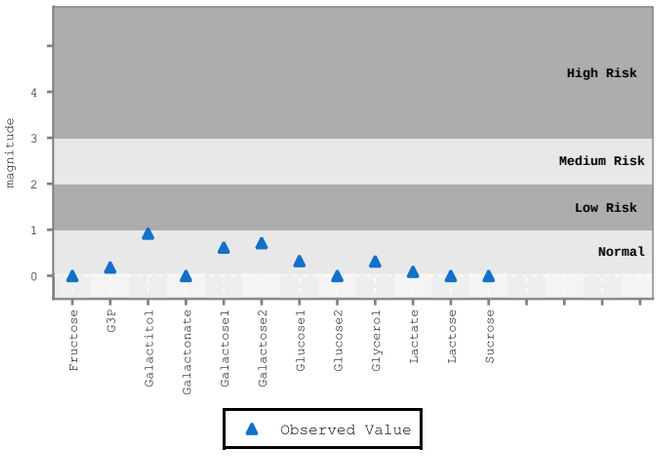


Sr. No.	Metabolite Name	Control Value	Observed Value
1	5HMU	0-0.1	N.D
2	DHT	0-0.9	N.D
3	DHU	0-1.5	N.D
4	hypoxanthine	0-4.2	N.D

Sr. No.	Metabolite Name	Control Value	Observed Value
5	Orotate	0-0.1	0.086
6	Thymine	0-0.2	0.195
7	Uracil	0-9.9	0.289

6. Disorders of Sugars:

Sr. No	Name of the Disorder	Result
1	Fructose-1 and 6-diphosphatase deficiency	Negative
2	Galactosemia	Negative
3	Endogenous sucrosuria	Negative
4	D-glyceric aciduria	Negative
5	Lactose Intolerance	Negative
6	Galactokinase deficiency (GALK)	Negative
7	Galactose epimerase deficiency (GALE)	Negative
8	Fructosuria	Negative
9	Transient Galactosemia	Negative

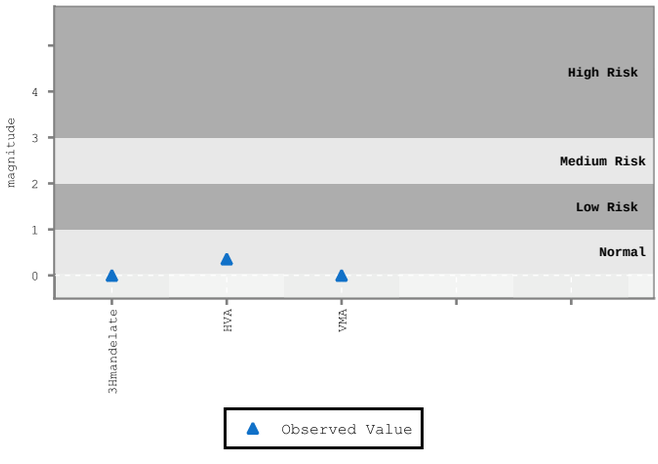


Sr. No.	Metabolite Name	Control Value	Observed Value
1	Fructose	0-1	N.D
2	G3P	0-4.2	0.773
3	Galactitol	0-7.7	7.092
4	Galactonate	0-10.6	N.D
5	Galactose1	0-0.4	0.25
6	Galactose2	0-1.4	1.031
7	Glucose1	0-35.3	11.459
8	Glucose2	0.4-27.5	N.D

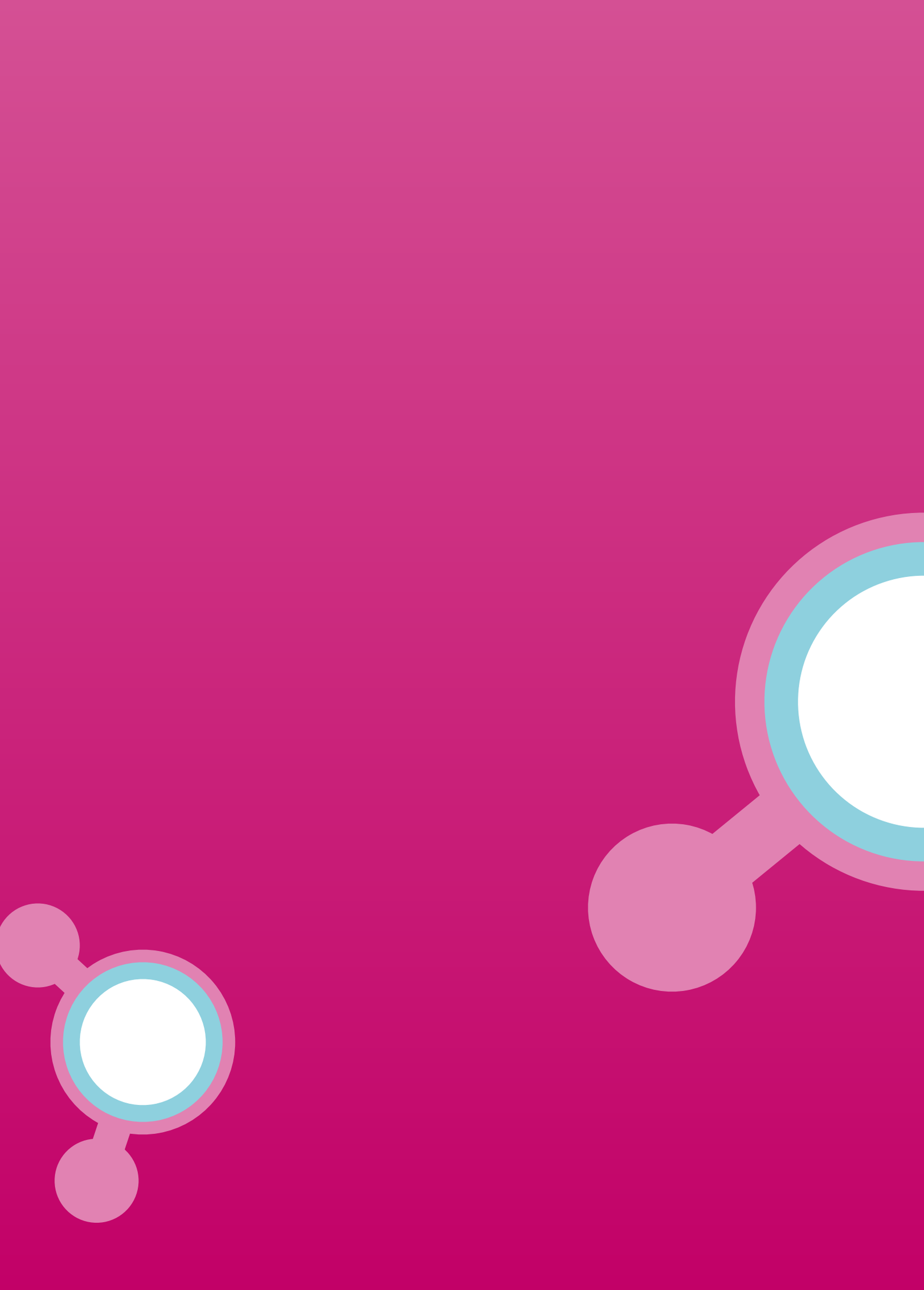
Sr. No.	Metabolite Name	Control Value	Observed Value
9	Glycerol	0-28.8	9.101
10	Lactate	1.1-208.1	19.121
11	Lactose	0.2-28	N.D
12	Sucrose	0-2.6	N.D

7. Non-IEM Disorder:

Sr. No	Name of the Disorder	Result
1	Neuroblastoma	Negative



Sr. No.	Metabolite Name	Control Value	Observed Value
1	3Hmandelate	0-0.1	N.D
2	HVA	0.1-1.6	0.565
3	VMA	0-6.5	N.D



SCIENCE

This chapter shows the science behind the test.

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All our analyses and treatment recommendations are scientifically validated. Here are some of the relevant literature references for your information.

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ADDITIONAL INFORMATION

In this chapter you will receive useful information

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ISO 22000:2018 No.00521/0

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Questions or comments about our service?

Our customer service team is happy to help with any enquiries or problems. You can contact us in the following ways:

- Phone +41 (0) 41 525 100.1
- office.ch@progenom.com

Our team is looking forward to your call. Customer satisfaction is our first priority. If you are not fully satisfied with our service, please let us know. We will do our best to help find a satisfactory solution to your problem.

Contact | Impressum
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Riedstrasse 1
6343 Rotkreuz
SWITZERLAND

Technical details

Order number
DEMO_ML

Date of birth
01/01/1990

Established analysis methods
qRT-PCR, DNA sequencing, fragment length analysis, CNV assay, GC-MS, Immunocap ISAC, Cytolisa

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Product codes
B1BAB, B2MIL, B4PRE

Current version
V538

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SWITZERLAND

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83395 Freilassing
Deutschland

Laboratory Director

Dr. Daniel Wallerstorfer Bsc.

Laboratory Manager

Florian Schneebauer, MSc.

NOTES:


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