

# Retrospective Study of Patients with Suspected Inborn Errors of Metabolism at Siriraj Hospital, Bangkok, Thailand (1997-2001)

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**Introduction:** This retrospective clinical study was carried out on patients with suspected inborn errors of metabolism (IEM) at Siriraj Hospital during 1997-2001. The authors investigated 114 patients by quantitative plasma amino acid analysis.

**Objective:** The objective of this study was to collect and analyze epidemiologic and specific clinical data of IEM, especially in small-molecule diseases.

**Material and Method:** All patients were categorized into 2 major groups: 1) positive diagnoses for IEM 2) negative diagnoses for IEM. The two groups were investigated, studied including statistical analysis.

**Results:** The authors found that most IEM ascertained through plasma amino acid analysis were small-molecule diseases (74.3%) and amino acid disorders consisted of the most frequent disorders. The presented data demonstrated that the ratio of positive diagnoses to all patients studied was 1:8. Epidemiological data showed there were more male than female patients. Onset of diseases occurred predominantly during the first month of age, and was rarely found after 3 years of age. There were histories of consanguinity in half of the IEM patients. The most common presenting symptom was acute metabolic encephalopathy and specific signs for small-molecule disorders included hepatomegaly, unusual urine odor, acidosis, hyperammonemia, alteration of consciousness, and ketosis/ketonuria. These signs or symptoms indicated further metabolic investigations.

**Conclusion:** Comparison of the data from Thailand with other countries showed both similarities and differences to the Caucasian population. Thus, further studies in IEM are much needed for the Thai population.

**Keywords:** Inborn errors of metabolism, Small-molecule diseases, Amino acid disorders, Consanguinity

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Inborn errors of metabolism (IEM) or inherited metabolic disorders are defined as a group of disorders that have different defects in the metabolic process, for example, enzymes, receptors, transport proteins, cofactors etc. But most biochemical geneticists usually define IEM as a group of disorders that have defects in catabolism or anabolism of nutrients

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or energy-producing molecules. They are caused by mutations in the genes that encode specific enzymes or cofactors<sup>(1,3)</sup>. Therefore, all IEM are genetically determined diseases that affect both patients and their families. Besides the treatment of individual patients, genetic counseling, including family support and prenatal diagnosis, are integral parts of the management of IEM patients. Multidisciplinary care teams including geneticists, pediatricians, nutritionists, biochemical scientists, developmental pediatricians and

other experts are required to take care of these patients and their families.

Sir Archibald Garrod discovered inborn errors of metabolism since the early 19th century. More than 350 different IEM have been described to date, and most of these are rare diseases/conditions<sup>(1)</sup>. Metabolic disorders account for a substantial percentage of the morbidity and mortality directly attributable to genetic diseases. A recent survey conservatively estimated that the incidence of metabolic disorders is approximately 1 in every 2,500 live births<sup>(1,22)</sup>, or 10% of all monogenic conditions in children. There has been a dramatic increase in understanding, novel diagnostic tests and treatment of these diseases in developed countries. However, studies of IEM are still in the early stage in Thailand and other developing countries. In the past, most cases were clinically diagnosed since there were few experts in metabolic diseases and lack of diagnostic facilities to provide the exact diagnosis. The prognoses of these patients were very poor due to the delay in diagnosis and lack of effective treatments. Most physicians in Thailand still consider IEM as very rare diseases which offer no treatment. Without governmental health-care insurance, most patients can not afford the expenses for laboratory diagnosis and treatment of these disorders. Special milk formulae and medications for these patients are still not included in the "30 baht health-care plan" governmental support.

In fact, the overall incidence of inherited metabolic disorders is not rare. The incidence in British Columbia, Canada was 1:2,500<sup>(13)</sup>, and in Italy was 1:2,555<sup>(14)</sup>. In Thailand, there has been no study on the exact incidence. From a multi-center study of IEM in Thailand<sup>(11)</sup> from 1988 to 2001, there were a cumulative total of 167 cases of all types of IEM, but this does not represent the total number of these patients in the Thai population. Some of the patients were diagnosed as having sepsis or other diseases, and may expire before referral to the medical schools to be taken care of by IEM experts.

The authors realize the importance of an epidemiologic study for this group of patients. The Division of Medical Genetics, Department of Pediatrics, Siriraj Hospital has been the pioneer in the study, research and care of these patients since 1989 and has collaborated with the Chulabhorn Research Institute since 1999. The objective of the present study was to collect the epidemiological and clinical information on all suspected cases and diagnosed cases of IEM for statistical analysis.

## Material and Method

The medical records of children suspected of or diagnosed with inborn errors of metabolism in the Department of Pediatrics, Siriraj Hospital, Mahidol University from 1997 to 2001 were retrospectively reviewed. All cases were investigated by metabolic work up, including quantitative plasma amino acid analysis, performed by high performance liquid chromatography (HPLC) in collaboration with the Chulabhorn Research Institute<sup>(18)</sup>.

From a total of 138 cases, 24 cases were excluded due to lack of medical records. Therefore, 114 patients were enrolled in the present study, who were divided into 2 groups:

1) Patients suspected of IEM: These patients were proven by the metabolic work up confirming that they were not IEM cases.

2) Patients diagnosed as having IEM: These patients were proven to be IEM cases.

Retrospective review of medical records gathered demographic data and clinical data. The demographic data included gender, ethnicity, age, and domicile. The clinical data included history of consanguinity, family history of affected cases or death in the infancy period with unknown cause in siblings, and clinical manifestations. The data from both groups was presented as pisdiaqram, bardigram and table as well.

All cases of IEM were further classified into the following subgroups: Amino acid disorders, organic acid disorders, fatty acid oxidation disorders, disorders of carbohydrate metabolism, lysosomal storage disorders, and disorders of metal transport.

## Statistical analysis

Comparison between cases and controls for categorical variables (e.g., gender, race, and consanguinity) were carried out using Chi-square test or Fisher's exact test (2-sided test). P-values of less than or equal to 0.05 were considered statistically significant. SPSS/PC Version10 was used for all statistical analyses.

## Results

From 1997 to 2001, there were 114 new patients who underwent metabolic work up, including plasma amino acid analysis. These patients comprised 35 cases (30.7%) of IEM, 41 cases (35.9%) of children with delayed development, and 38 cases (33.33%) cases of non-IEM diseases (e.g. kernicterus, congenital infections, perinatal asphyxia, cerebral palsy, cerebral

dysgenesis, Reye syndrome etc.). Of the 35 cases of IEM, 26 cases (22.8%) were confirmed by plasma amino acid analysis, urine organic acid analysis or tandem mass spectrometry, and 9 cases (7.9%) were only clinically diagnosed, because specific tests (e.g. enzyme assays) were not available.

All IEM cases found are shown in Fig. 1. The IEM cases were divided into 6 groups as follows: Amino acid disorders, organic acid disorders, fatty acid oxidation defect disorders, disorders of carbohydrate metabolism, lysosomal storage diseases, and disorders of metal transport. Seventeen cases of amino acid disorders consisted of 6 cases of maple syrup urine disease, 4 cases of urea cycle defects (2 cases of ornithine transcarbamylase deficiency, 1 case of argininosuccinate synthetase deficiency, 1 case of argininosuccinate lyase deficiency), 2 cases of non-ketotic hyperglycinemia, 2 cases of homocystinuria, 1 case of phenylketonuria, 1 case of (suspected) tyrosinemia type I, and 1 case of oculocutaneous albinism. Five cases of organic disorders consisted of 2 cases of isovaleric acidemia, 2 cases of methylmalonic acidemia, and 1 case of multiple carboxylase deficiency.

Three cases of fatty acid oxidation defects consisted of 1 case of short-chain acyl CoA dehydrogenase deficiency, 1 case of primary carnitine deficiency, and 1 case of medium-chain acyl CoA dehydrogenase deficiency. One case of metal transport defect was Menkes disease. Among lysosomal storage disorders were 7 cases of leukodystrophy and 1 case of I-cell disease.

Gender, age of onset, history of consanguinity were compared between the 2 groups of

**Table 1.** Comparison of gender, age of onset, history of consanguinity between Non-IEM and IEM cases

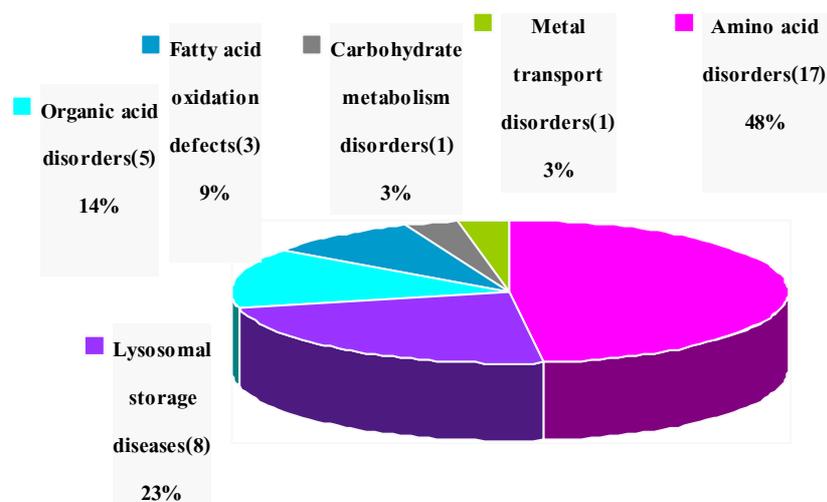
	Non-IEM (n = 79)	IEM (n = 35)	p-value
1) Gender			
Male	34 (43.0%)	23 (65.7%)	0.026*
Female	45 (57.0%)	12 (34.3%)	
2) Age of onset			
< 1 month	29 (39.7%)	22 (62.9%)	0.024*
> 1 month	73 (60.3%)	35 (37.1%)	
3) History of consanguinity			
No	73 (92.4%)	18 (51.4%)	<0.001*
Yes	6 (7.6%)	17 (48.6%)	

\* Chi-squared test

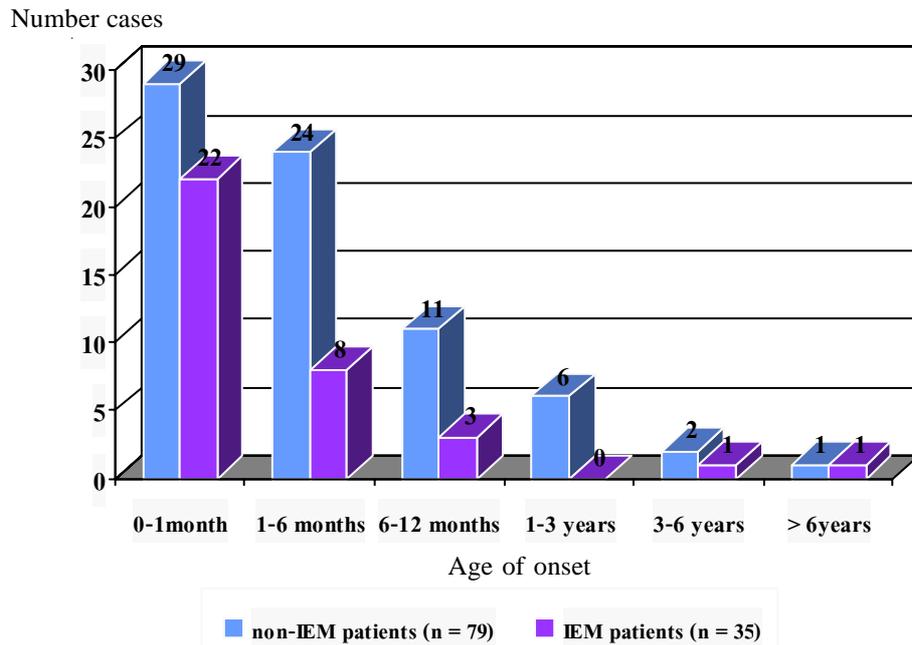
patients, IEM cases and non-IEM cases, as shown in Fig. 2 and Table 1.

The median age of onset was calculated to be 7.3 days (minimum = day 1, maximum = 9 years) in the IEM group and 3 months (minimum = day 1, maximum = 11 years) in the non-IEM group. Thus, the majority of IEM cases had onset in the neonatal period, while the majority of the non-IEM group had onset beyond the neonatal period. From Table 1, M:F ratio in the IEM group was 1.9:1 and in the non-IEM group was 1:1.3, indicating a significant male preference in the IEM group. In terms of consanguinity history, there were consanguineous marriages in nearly 50% of cases with IEM, but very few in non-IEM cases.

The presenting symptoms, signs and laboratory results were also compared between IEM and non-IEM groups as shown in Tables 2, 3, and 4.



**Fig. 1** Diagram shows 6 groups of IEM cases found (n = 35)



**Fig. 2** Comparison between age of onset of IEM cases and non-IEM cases

From Table 2, 48.6% of IEM cases had presenting symptoms of acute metabolic encephalopathy (e.g. coma, cerebral edema), which was the leading symptom in the IEM groups. On the contrary, with the non-IEM group, presentation with developmental delay was found in 51.9% of cases. From Table 3, the most common associated sign for IEM group was hepatomegaly, which was significantly different when compared with the non-IEM group. Unusual urine odor and dysmorphism were also significantly more frequent in the non-IEM group than in the IEM group. Other signs did not differ significantly between the two groups. From Table 4,

metabolic acidosis, hyperammonemia, and ketosis were significantly more frequently found in the IEM group than in the non-IEM group. However, frequency of hypoglycemia, lactic acidosis and cytopenia were not significantly different between the two groups.

### Discussion

Inborn errors of metabolism (IEM) are a group of diseases that require biochemical workup, such as plasma amino acid analysis, urine organic acid analysis and tandem mass spectrometry. But some disorders such as lysosomal storage disorders need advanced facilities to diagnose, including leukocyte

**Table 2.** Presenting symptoms of non-IEM and IEM groups

Presenting symptoms	Non-IEM (n = 79)	IEM (n = 35)	p-value
Acute metabolic encephalopathy	5 (6.3%)	17 (48.6%)	<0.001*
Developmental delay	41 (51.9%)	9 (25.7%)	0.009*
Seizure	13 (16.5%)	4 (11.4%)	
Metabolic acidosis	4 (5.1%)		
Ocular symptoms	2 (2.5%)	2 (5.7%)	
Hepatic symptoms		1 (2.9%)	
Heart failure/muscle weakness	1 (1.3%)	1 (2.9%)	
Cerebral palsy	6 (7.6%)		
Others	7 (8.9%)	1 (2.9%)	

\* Chi-squared test

**Table 3.** Associated signs of non-IEM and IEM groups

Associated signs	Non-IEM (n = 75)	IEM (n = 35)	p-value
Hepatomegaly	4 (5.1%)	11 (31.4%)	<0.001*
Ocular abnormalities	20 (25.3%)	9 (25.7%)	0.964
Unusual urine odor	2 (2.5%)	6 (17.1%)	0.010*
Hypopigmentation	5 (6.3%)	3 (8.6%)	0.699
Cardiomyopathy/Myopathy	3 (3.8%)	1 (2.9%)	1.000
Dysmorphism	13 (16.5%)	1 (2.9%)	0.009*

\*Fisher exact test

**Table 4.** Laboratory results of non-IEM and IEM groups

Lab results	Non-IEM (n = 75)	IEM (n = 35)	p-value
Metabolic acidosis	19 (12.4%)	15 (42.9%)	0.043*
Hyperammonemia	1 (1.3%)	12 (34.3%)	<0.001*
Ketosis	3 (3.8%)	7 (20.0%)	0.009*
Hypoglycemia	5 (6.3%)	3 (8.6%)	0.699
Lactic acidosis	7 (8.9%)	3 (8.6%)	1.000
Cytopenia	2 (2.6%)	3 (8.6%)	0.168

\* Chi-squared test/Fisher exact test

or fibroblast enzyme assay. At that time of the present study, there was no HPLC and GC/MS in the Division of Medical Genetics, Department of Pediatrics, Siriraj Hospital; so the authors collaborated with other institutes both within the country and overseas. The Chulabhorn Research Institute was one of the institutes that offered plasma amino acid analysis for the presented patients, helping in the diagnosis of several amino acid disorders. From the present study, there were 18 cases showing abnormal results on plasma amino acids analysis. When compared with all cases (138 cases) analysed, the ratio was 1:8. To the best of the authors' knowledge, there is no previous data showing the ratio between positive cases to all cases investigated by plasma amino acids analysis in Thailand.

From the present study, 74% of IEM cases were small-molecule diseases such as amino acid disorders, organic acid disorders, fatty acid oxidation disorders, and carbohydrate metabolism (gluconeogenesis) disorders. Twenty-three percent of cases were lysosomal storage disorders (most cases were leukodystrophies). Because plasma amino acid analysis was not part of the investigation for the patients with suspected macromolecule diseases (lysosomal storage disorders); thus cases of lysosomal storage disorders were not included in the present study. This may be compared to the incidence of IEM in developed countries, where half of all IEM cases were small molecule diseases<sup>(1,14,22)</sup>.

When all cases of small-molecule diseases were analysed, 65% were amino acid disorders (urea cycle defect included). This is comparable with the data from other studies<sup>(13,14)</sup>. From a multi-center study in Thailand, amino acid disorders and urea cycle defect cases comprised of 74.3% of all small-molecule diseases cases<sup>(11)</sup>. Italy and Canada showed 38.3% and 78.6% of cases respectively<sup>(13,14)</sup>. Half of the amino acid disorders cases in Western countries were phenylketonuria (PKU) which has an incidence of 1:10,000 live births<sup>(1)</sup>. But the true incidence of PKU in Thailand is as yet unknown.

Epidemiological data from the present study showed that there were more male than female patients by a ratio of 2:1. This may be due to the fact that some IEMs are transmitted as X-linked inheritance. Menkes disease and ornithine transcarbamylase deficiency, found in the present study, are X-linked recessive. The authors found that most IEM cases were manifested early in the neonatal period. The remaining were insidious and chronic progressive diseases, such as PKU or homocystinuria. Study of IEM in Italy showed similar findings regarding age of onset, with 75% of small-molecule disease cases having onset before 3 years of age<sup>(14)</sup>.

Consanguineous marriages are more likely to produce offspring affected by autosomal recessive disorders because relatives more often share disease genes inherited from a common ancestor. Most IEM

are autosomal recessive disorders, and the authors found that 50% of IEM cases in the present study had a history of consanguinity.

About half of IEM patients in the present study had acute encephalopathy as the most frequent presenting symptom. All cases that presented with this symptom were small-molecule disorders. The toxic metabolites that accumulate in plasma diffuse through the blood brain barrier and enter into neurons and supporting cells, causing cellular swelling and dysfunction. Other symptoms, specific for IEM in the present study, were enlarged liver, abnormal urine odor, metabolic acidosis, hyperammonemia, and ketosis. Ketonuria in the sick newborn strongly suggests metabolic disorders in the infant. Increased plasma lactate can be found in metabolic disorders of oxidative phosphorylation, Krebs's cycle, and gluconeogenesis. However, it can increase in other conditions such as shock or technical errors in blood drawing.

Thus, patients with history of consanguinity, having onset of symptoms and acute encephalopathy within the first month of life, should undergo thorough investigations for metabolic disorders. However, patients without such a history or symptoms should not be excluded from differential diagnosis of IEM, due to variability of symptoms and severity and non-specificity of IEM.

### Conclusion

The present study provides clinical data of IEM patients who were investigated at the Division of Medical Genetics, Department of Pediatrics, Siriraj Hospital Faculty of Medicine, in collaboration with the Chulabhorn Research Institute, prior to the establishment of the genetic metabolic center at the Division of Medical Genetics, Department of Pediatrics, Siriraj Hospital Faculty of Medicine in 2001. Epidemiological comparison of data, between the present study and previous studies from both domestic and overseas, showed some similarities. The authors were able to provide biochemical investigations to the presented patients and referrals from other hospital. However, further studies of IEM are required in Thailand. The authors' goal is to make general practitioners and pediatricians realize the importance of early diagnosis and treatment of IEM.

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## การศึกษาย้อนหลังในผู้ป่วยที่สงสัยโรคพันธุกรรมเมตาบอลิก ณ โรงพยาบาลศิริราช (2540-2544)

พรสวรรค์ วสันต์, นิธิวัชร วัฒนวิจารณ์, จันทรกานต์ ศรีสมทรัพย์, พรรณี สว่างอารีตระกูล, สมพร เหลี่ยมมงคลกุล, ชีษณสร สวัสดิวัฒน์

**บทนำ:** การศึกษานี้เป็นการศึกษาย้อนหลังในผู้ป่วยที่สงสัยโรคพันธุกรรมเมตาบอลิก ณ โรงพยาบาลศิริราช ระหว่างปีพ.ศ. 2540-2544 และได้ทำการศึกษาผู้ป่วยทั้งหมด 114 รายโดย การตรวจวิเคราะห์กรดอะมิโน ในเชิงปริมาณ

**วัตถุประสงค์:** เพื่อรวบรวมและวิเคราะห์ทางระบาดวิทยา รวมทั้งข้อมูลทางคลินิกของผู้ป่วยโรคพันธุกรรมเมตาบอลิก โดยเฉพาะอย่างยิ่งในโรคพันธุกรรมเมตาบอลิกที่เกิดจากสารโมเลกุลเล็ก

**วัสดุและวิธีการ:** แยกผู้ป่วยทั้งหมดเป็น 2 กลุ่ม กลุ่มแรกได้รับการวินิจฉัยเป็นโรคพันธุกรรมเมตาบอลิก กลุ่มที่ 2 ไม่ได้รับการวินิจฉัยเป็นโรคพันธุกรรมเมตาบอลิก ทั้ง 2 กลุ่มนี้ได้รับการศึกษาวิเคราะห์โดยมีนัยสำคัญในทางสถิติ

**ผลการศึกษา:** จากผู้ป่วยที่ได้รับการยืนยันว่าเป็นโรคพันธุกรรมเมตาบอลิกจริง โดยการ ตรวจวิเคราะห์ ปริมาณของกรดอะมิโน พบว่าร้อยละ 74.3 เกิดจากสารโมเลกุลเล็ก และความผิดปกติของกรดอะมิโนเป็นโรคที่พบได้บ่อยที่สุด ผลการศึกษายังแสดงให้เห็นสัดส่วนของผู้ป่วยที่การวินิจฉัยโรคที่ให้ผลบวกต่อจำนวนผู้ป่วยทั้งหมดเท่ากับ 1 ต่อ 8 ข้อมูลทางระบาดวิทยาแสดงให้เห็นว่าพบเพศชายมากกว่าเพศหญิง อายุที่เกิดโรคมักพบได้บ่อยในเดือนแรกของชีวิต และพบน้อยภายหลังจากอายุ 3 ปี พบประวัติการแต่งงานในเครือญาติถึงร้อยละ 50 ในผู้ป่วยพันธุกรรมเมตาบอลิก อาการที่พบบ่อยที่สุดได้แก่ อาการทางสมองเฉียบพลัน (acute metabolic encephalopathy) รวมทั้งภาวะโคม่า และอาการแสดงที่จำเพาะสำหรับโรคที่เกิดจากสารโมเลกุลเล็ก ได้แก่ ตับโต กลิ่นปัสสาวะแปลก ภาวะกรดคั่งในเลือด ภาวะแอมโมเนียคั่งในเลือดการเปลี่ยนแปลงของสติสัมปชัญญะ และภาวะคีโตนคั่งในเลือดและปัสสาวะ อาการและอาการแสดงทั้งหมดนี้บ่งชี้ว่าแพทย์ควรส่งตรวจเพิ่มเติมทางเมตาบอลิก

**สรุป:** การเปรียบเทียบข้อมูลของผู้ป่วยโรคพันธุกรรมเมตาบอลิกในประเทศไทยกับต่างประเทศ พบทั้งความคล้ายคลึงและความแตกต่างกัน ดังนั้นการศึกษาเพิ่มเติมในผู้ป่วยพันธุกรรมเมตาบอลิกในประเทศไทย จึงเป็นสิ่งจำเป็นอย่างยิ่ง

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