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SCIENTIFIC LETTER

Patients with inborn errors of metabolism in an emergency department

Pacientes con enfermedad por error congénito del metabolismo visitados en un servicio de urgencias

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Inborn errors of metabolism (IEMs) are a heterogeneous group of low-incidence diseases caused by mutations in genes encoding enzymes or other functional proteins. These mutations lead to the accumulation of unmetabolized substrates and a deficiency of the corresponding products, which underlies the clinical signs and symptoms of the disease, some of which can be life-threatening[1,2]. In our region, diagnosis is primarily made through the neonatal screening program and subsequently confirmed by genetic testing[3]. Some affected patients require specialized follow-up by the Nutrition and Dietetics Department and specific ongoing treatment, including medication and/or dietary intervention[4]. However, when they develop an intercurrent illness that may trigger metabolic decompensation, they seek care in the Pediatric Emergency Departments (PEDs). These patients require specific management and treatment[5], with which emergency pediatricians have limited familiarity: given the low prevalence of certain disorders, clinical experience is limited, making it difficult to recognize characteristic clinical patterns and initiate specific pharmacological and dietary treatments. The aim of our study is to determine how frequently patients with IEMs seek care in a PED, as well as the most common reasons for consultation and types of metabolic decompensation.

We present a retrospective, descriptive-observational study conducted in the emergency department (ED) of a tertiary maternal and child hospital in Spain. Patients under 18 years of age diagnosed with an inborn error of metabolism (IEM) between 2017 and 2022 and followed by the Nutrition and Dietetics Department were included. Patients diagnosed with more than one IEM and/or lost to follow-up were excluded. IEMs were classified as amino acid disorders (IEM-aa), organic acid disorders (IEM-OA), and fatty acid disorders (IEM-FA). ED reports from the two years following diagnosis were reviewed, and clinical-epidemiological and management variables were collected. For all patients, the triage level assigned before medical assessment was recorded using the Andorran Triage Model, along with the Pediatric Assessment Triangle (PAT) and the Gorelick dehydration scale. Metabolic decompensation was defined as the presence of one

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or more of the following laboratory abnormalities: hypoglycemia, metabolic acidosis, hyperlactatemia, hyperammonemia, and elevated transaminases. Data were analyzed using IBM® SPSS® Statistics for Windows® version 28. The study was approved by the Ethics Committee of the study hospital (PIC-158-24).

During the study period, 87 patients were diagnosed with an IEM; 10 were excluded (8 due to loss to follow-up at the study center and 2 with two IEMs). The final sample comprised 77 patients, with a breakdown by IEM type and frequency shown in Table 1. Forty-three (55.8%) were male, and the median age at presentation was 15 days (range 3 days–6.5 years).

TABLE 1.

Type of inborn error of metabolism (n=77).

Amino acid disorders	**32**
Phenylketonuria	21
Homocystinuria	4
Tyrosinemia I	3
Hypermethioninemia	2
Maple syrup urine disease	1
Citrullinemia	1
Organic acid disorders	**24**
3-Methylcrotonylglycinuria	11
Glutaric aciduria type I	4
Propionic acidemia	3
Methylmalonic acidemia	3
Argininosuccinic aciduria	1
Isovaleric acidemia	1
3-Hydroxy-3-methylglutaryl-CoA lyase deficiency	1
Fatty acid metabolism disorders	**21**
Medium-chain acyl-CoA dehydrogenase deficiency	9
Very long-chain acyl-CoA dehydrogenase deficiency	6
Short-chain acyl-CoA dehydrogenase deficiency	5
Cellular carnitine uptake deficiency	1

Thirty-five patients (45.45%) presented to the PED during the two years following disease onset (10 [31.3%] with an IEM-aa, 12 [50%] with an IEM-OA, and 13 [61.9%] with an IEM-FA; *p=0.078*), accounting for 155 visits, with a median of 3 visits per patient (p25–75 1–6), with no differences by type of metabolic disorder (*p=0.487*).

Table 2 shows the characteristics of these consultations. Patients presented with an abnormal PAT in 8 (5.2%) visits, all due to respiratory distress. Metabolic decompensation was diagnosed in 20 (12.9%) consultations, all in clinically normohydrated patients (Gorelick scale score 0). These decompensations affected 12 patients (9 decompensations in 5 patients with an IEM-OA and 11 in 7 patients with an IEM-FA). In order of frequency, the following metabolic abnormalities were detected: hyperlactatemia, 12; metabolic acidosis, 9; elevated transaminases, 7; hyperammonemia, 6; and hypoglycemia, 2 (11 consultations involved more than one abnormality). Fifteen of the 20 decompensations occurred in the context of gastroenteritis and 5 in the context of respiratory infection. Hospital admission was recommended in 15 cases, none to an intensive care unit (ICU).

Most IEM diagnoses were made through the neonatal screening program, explaining why the median age at diagnosis fell within the neonatal period[1]. Diagnoses made outside this period occurred mainly in patients who had not undergone neonatal screening. This study shows that the reasons for presentation and diagnoses in the ED among patients with IEM are similar to those in the general pediatric population, although their management differs[6]. The most common group is amino acid metabolism disorders (AAMDs)[1], and its members attend the ED least often. AAMDs do not usually present with acute decompensations, but rather with neurological symptoms in the medium and long term, such as seizures, myoclonus, and psychomotor delay, due to the toxic effects of amino acid accumulation in different tissues[7,8]. Notably, in this study, these patients were triaged as urgent (triage levels II and III), and in 4 cases, blood tests including a metabolic profile were performed during the medical consultation, even though the patients were stable and not dehydrated and their risk of acute metabolic decompensation was relatively low. This may reflect the emergency department staff's limited experience in the clinical management of this group of disorders.

Organic acidemias and fatty acid disorders are less common, but patients with these conditions seek care more often and, when they do, have a considerable rate of acute metabolic decompensation. Gastroenteritis is the main cause of decompensation, since prolonged fasting and stress can trigger it[6,9]. These patients have a very high admission rate because of the instability associated with the underlying metabolic disorder[10,11,12]. In other studies[13,14], these groups required intensive care admissions for neurological symptoms; however, in our study, no patient with decompensation presented with this alteration.

The limitations of the study include those inherent to its retrospective design. Furthermore, this was a single-center study conducted at a tertiary maternal and child hospital, so the results may not be generalizable to healthcare centers with different characteristics. Finally, the small number of cases included, together with the fact that these patients have specific home-based recommendations in the event of suspected metabolic decompensation, makes it difficult to identify risk factors associated with decompensation.

TABLE 2. Characteristics of emergency department visits by type of inborn error of metabolism (n = 155)

	IEM-aa (n=27)	IEM-OA (n=63)	IEM-FA (n=65)	*p*
Triage II-III	17 (63%)	57 (90.5%)	47 (72.3%)	***0.005***
Abnormal PAT	0 (0%)	4 (6.3%)	4 (6.2%)	*0.410*
Reason for visit **Fever** **Gastrointestinal** **Respiratory** **Other**	11 (40.7%) 4 (14.8%) 4 (14.8%) 8 (29.6%)	20 (31.7%) 19 (30.2%) 11 (17.5%) 13 (20.6%)	29 (44.6%) 17 (26.2%) 12 (18.5%) 7 (10.8%)	*0.283*
MET laboratory tests **Not performed** **No abnormalities** **Abnormal**	23 (85.8%) 4 (14.8%) 0 (0%)	22 (34.9%) 30 (47.6%) 11 (17.5%)	42 (64.6%) 14 (21.5%) 9 (13.8%)	***<0.001***
Diagnosis **Fever** **URTI** **AGE** **LRTI** **Other**	6 (22.2%) 8 (29.6%) 3 (11.1%) 2 (7.4%) 8 (29.6%)	11 (17.5%) 15 (23.8%) 18 (28.6%) 5 (7.9%) 14 (22.2%)	21 (32.3%) 10 (15.4%) 12 (18.5%) 9 (13.8%) 13 (20%)	*0.241*
Admission	3 (11.1%)	21 (33.3%)	11 (16.9%)	***0.025***

IEM: inborn errors of metabolism. Amino acid IEMs: inborn errors of amino acid metabolism. Organic acid IEMs: inborn errors of organic acid metabolism. Fatty acid IEMs: inborn errors of fatty acid metabolism. PAT: Pediatric Assessment Triangle. MET laboratory tests: laboratory tests with a metabolic profile. URTI: upper respiratory tract infection. AGE: acute gastroenteritis. LRTI: lower respiratory tract infection.

In conclusion, acute metabolic decompensations occur in patients with IEM-OA and IEM-FA. In contrast, patients with IEM-aa have a low risk of decompensation, and routine blood tests should be avoided to minimize discomfort, costs, and potential false-positive results that could complicate patient management. Developing a checklist according to the type of IEM should be considered as a future strategy to individualize care for these patients in the ED.

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Conflict of interests

I declare that there is no commercial or financial conflict of interest related to this research.

Author Contributions

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Use of AI

I declare that no artificial intelligence tools (such as ChatGPT, Copilot, Gemini, or others) were used in the writing, analysis, or review of this article.

Data availability

The data supporting the results of this study are available upon request from the corresponding author.

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