Table (1): Distribution of studied neonates as per tandem mass spectrometry findings

Parameters			Studied neonates (n= 50)	
			n	%
TMS	No specific findings (Normal)		39	78.0%
	Abnormal (n= 11)	High leucine,isoleucine, valine, leucine:phenylalanine ratio and leucine:alanine ratio	2	4.0%
		High free carnitine	1	2.0%
		High propionyl carnitine (C3), and C3:C2 ratio	1	2.0%
		MCAD deficiency (High C6,C8,C10)	1	2.0%
		High citrulline and methionine	ort1	2.0%
		High C16-Carnitine , C6-Carnitine , C5-DC, C4-OH (C3-DC) ,and high C3 DC	4	8.0%
		High tyrosine level	1	2.0%

n: number, %: percentage, IEM: Inborn errors of metabolism, TMS: tandem mass spectrometry, MCAD: Medium-Chain Acyl-CoA Dehydrogenase Deficiency

Table (2): Distribution of studied neonates as per IEM disorders

	Parameters	Studied neonates (n= 50)	
		n	%
IEM	Normal screening for IEM	32	64.0%
	Urea cycle defect	7	14.0%
	MSUD	2	4.0%
	Methyl melanoic acidemia	1	2.0%
	Fatty acid oxidation defect	1	2.0%
	Confirmed organic academia	4	8.0%
	Confirmed tyrosinemia	1	2.0%
	Confirmed mitochondrial disorder	2	4.0%

n: number, %: percentage, IEM: Inborn errors of metabolism, MSUD: Maple syrup urine disease.

Table (3): Comparison between neonates with and without IEM cases regarding demographic and clinical data

Variable		Neonates without IEM (N=32)		Neonates with IEM (N=18)		Test value	P-value
		No.	%	No.	%		
Age (days)	Mean± SD	13.19±6.91		15.17±5.36		T= 1.05	0.300
	Median	13.0		16.0			
	Range	3.0- 25.0		4.0- 25.0			
Gestational Age (weeks)	Mean± SD	37.97± 1.12		36.2± 2.25		Z_{MWU}= 0.628	0.530
	Median	38.0		36.0			
	Range	32.0-39.0		32.0-39.0			
Weight (Kg)	Mean± SD	2.54± 0.75		2.88± 0.62		T= 1.93	0.110
	Median	2.56		3.06			
	Range	1.26- 3.67		1.49- 3.62			
Head circumference (cm)	Mean± SD	34.5± 2.54		34.25± 8.25		^{Z}MWU= 0.097	0.923
	Median	34.50		33.0			
	Range	29.0-36.0		29.0-36.0			
Gender	Male	25	78.1%	15	83.3%	X^2= 0.195	0.659
	Female	7	21.9%	3	16.7%		
Consanguinity	No	21	65.6%	8	44.4%	X^2= 2.12	0.145
	Yes	11	34.4%	10	55.6%		
Maturity	Full term	26	81.3%	15	83.3%	X^2= 0.0.34	0.854
	Preterm	6	18.8%	3	16.7%		
Presenting symptoms	Jaundice	0	0.0%	1	5.6%	X^2= 0.271	1.00
	Hepatomegaly	0	0.0%	1	5.6%	X^2= 0.271	1.00
	Vomiting	0	0.0%	7	38.9%	X^2= 11.42	**0.001**
	Convulsions	3	9.4%	8	44.4%	X^2= 6.34	**0.012**
	previous siblings' death	2	6.3%	1	5.6%	X^2= 0.271	0.602
	Persistent metabolic acidosis	3	9.4%	6	33.3%	X^2= 3.004	0.083
Neurological examination	Normal	15	46.9%	2	11.1%	X^2= 5.07	**0.024**
	Abnormal	17	53.1%	16	88.9%		

P value< 0.05 is significant, P value< 0.01 is highly significant, SD: Standard deviation, T= Student T test ZMWU = Mann- Whitney U test, X^2= Chi- Square test

Table (4): Comparison between neonates with and without IEM cases regarding laboratory data

Variable		Neonates without IEM (N=32)	Neonates with IEM (N=18)	Test value	P-value
Hb (gm %)	**Mean± SD**	18.8± 0.9	18.3± 0.7	Z_{MWU} = 1.04	0.341
	Range	15.2- 19.2	16.9 – 19.3		
WBCs ($cm^3 \times 10^9$/L)	**Mean± SD**	16.4± 7.9	14.1± 4.7	T= 1.39	0.172
	Range	2.7- 28.0	4.5- 21.5		
Platelets ($\times 10^3$/mcL)	**Mean± SD**	213.2± 52.5	227.4± 61.6	T= 0.966	0.338
	Range	84.0 - 347.0	85.0- 376.0		
AST	**Mean± SD**	31.25± 13.68	27.75± 5.28	Z_{MWU} = 1.56	0.118
	Range	17.0 - 140.0	21.0 - 38.0		
ALT	**Mean± SD**	17.06± 8.77	14.60± 6.82	Z_{MWU} = 1.08	0.279
	Range	5.0 – 70.0	6.0 – 26.0		
Urea	**Mean± SD**	14.93± 2.51	14.15± 2.5	Z_{MWU} = 1.29	0.197
	Range	10.0 – 19.0	11.0 – 19.0		
Creatinine	**Mean± SD**	0.72± 0.15	1.10± 1.96	Z_{MWU} = 0.758	0.449
	Range	0.30 – 1.00	0.30 – 9.40		
Serum ammonia	**Mean± SD**	40.43± 40.75	207.1± 117.9	Z_{MWU}= 2.69	0.007
	Range	3.0- 98.0	9.0- 313.0		
Serum lactate	**Mean± SD**	23.26± 3.88	30.31± 21.41	Z_{MWU}= 0.143	0.886
	Range	15.0- 30.0	15.0- 78.50		
ABG with anion gap	**Normal**	17 53.1%	9 50.0%	X^2= 4.98	0.083
	Acidosis with high anion gap	2 6.3%	5 27.8%		
	Acidosis	13 40.6%	4 22.2%		
Organic acid in the urine	**Not done**	31 96.9%	12 66.7%	X^2= 8.98	0.030
	Normal	1 3.1%	4 22.2%		
	High methyl malonic acid	0 0.0%	1 5.6%		
	Non-glucose reducing substance and G-1-P	0 0.0%	1 5.6%		

P value< 0.05 is significant, P value< 0.01 is highly significant, SD: Standard deviation, T= Student T test ZMWU = Mann- Whitney U test, X^2= Chi- Square test

Table (5): Distribution of studied neonates as per outcome

Parameters		Studied neonates (n= 50)	
		n	%
Outcome	**Discharged from NICU for follow up**	39	78.0%
	Died	8	16.0%
	Discharged as requested by parents and lost follow up	3	6.0%

n: number, %: percentage,

Table (6): Distribution of investigations of inborn errors of metabolism of the studied neonates

Parameters		Studied neonates	
		n	%
Serum ammonia (mg/dL) (n= 21/50)	**Mean± SD**	151.57± 126.6	
	Median	98.0	
	Range	3.0-313.0	
serum lactate (mg/dL) (n= 21/50)	**Mean± SD**	25.34± 12.21	
	Median	24.0	
	Range	15.0-78.5	
ABG with anion gap (n=50)	**Normal**	26	52.0%
	acidosis with high anion gap	7	14.0%
	Acidosis	17	34.0%
Organic acid in the urine (n= 7/50)	**Not done**	43	86.0%
	Normal	5	10.0%
	high methyl malonic acid	1	2.0%
	non-glucose reducing substance and galactose 1 phosphate	1	2.0%

Table (7): Validity of TMS in diagnosis of IEM in studied neonates

Item	IEM Diagnosis by Clinical finding, other modalities confirmed			
	No (n = 32)		**Yes (n = 18)**	
	n	**%**	**n**	**%**
IEM Diagnosis by TMS				
No (n= 39)	32	0%	7	9.5%
Yes (n= 11)	0	100%	11	90.5%