Al Jawhara Centre for Molecular Medicine & Inherited Disorders

مركز الجوهرة للطب الجزيئي وعلوم المورثات والأمراض الوراثية

Selective newborn screening of amino acid, fatty acid and organic acid disorders in the Kingdom of Bahrain.

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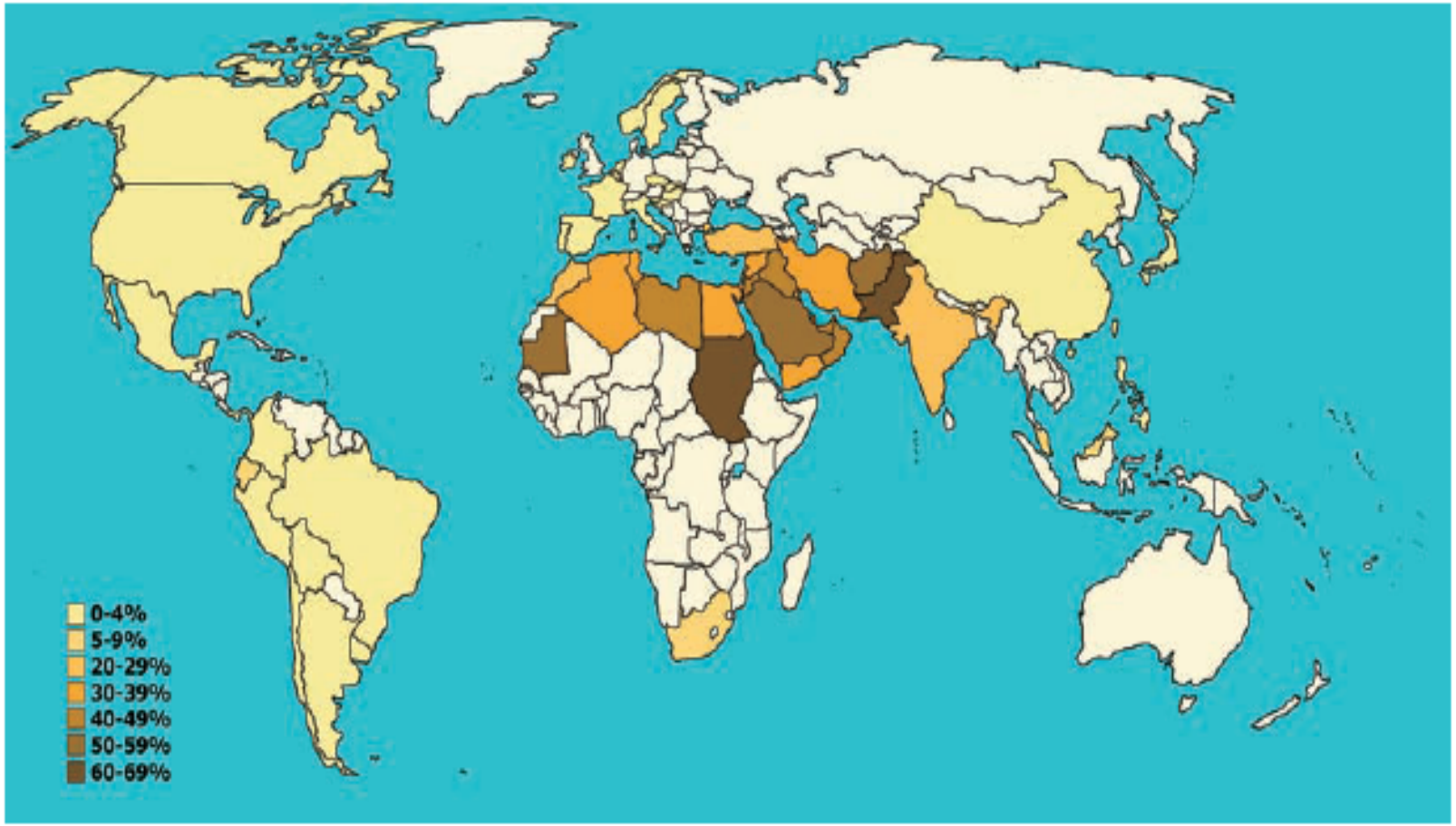
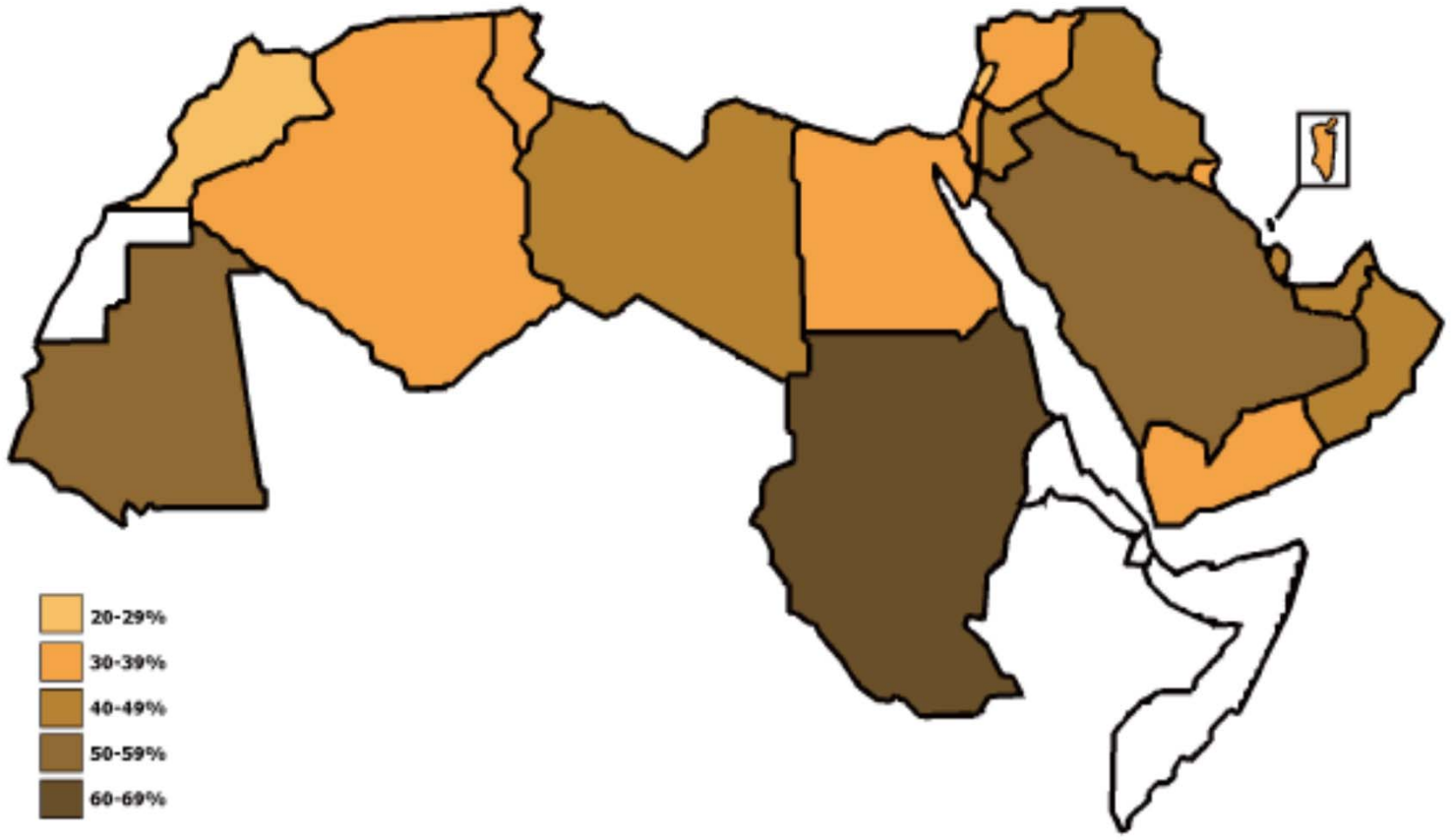
- Consanguinity rates in Arabian Peninsula
- NBS Programs In The Region
- Retrospective Study in Bahrain
- Conclusion

- Newborn Screening is **NOT WELL yet recognized** as an essential, preventive public health program in the region

- Some preliminary studies in the region showed the **incidences of these disorders are to be higher in the Middle East than anywhere else in the world** due to the **consanguinity**

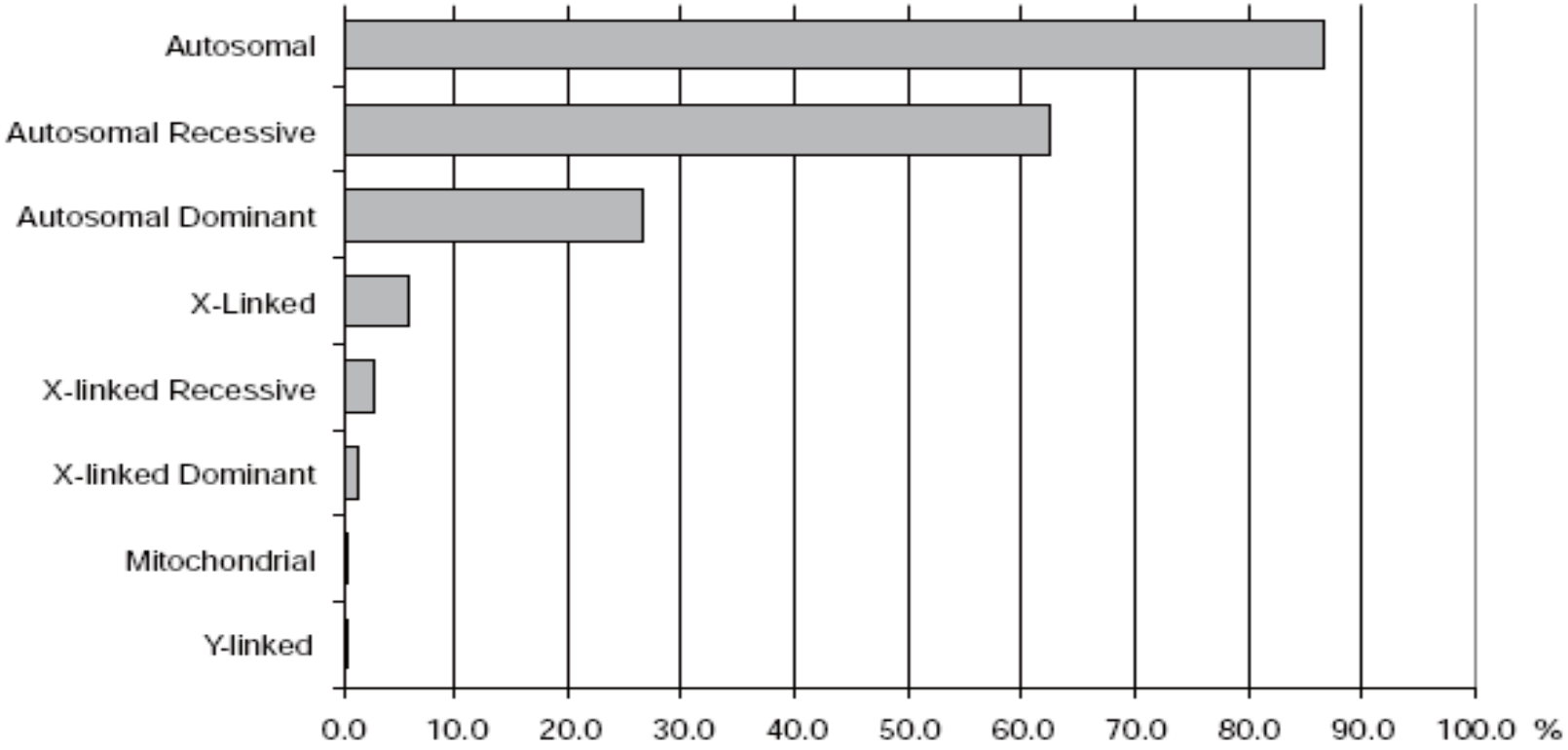
Consanguinity



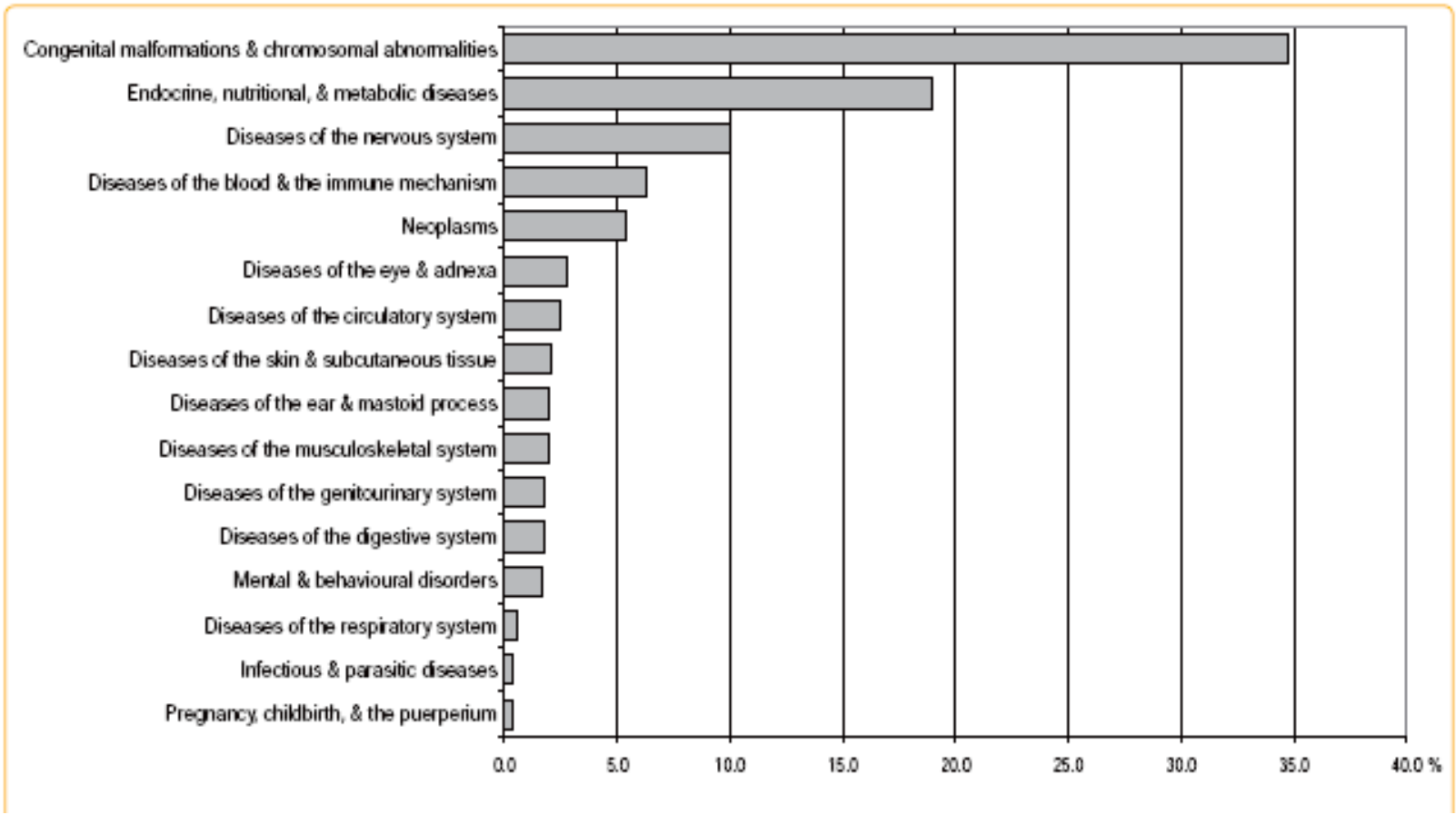


In the Arabian Peninsula, there are high percentages of consanguineous marriages and the tribal nature of marriages

Genetic Disorders



Classification of Genetic Disorders in Arabs according to mode of inheritance (WHO ICD-10, 2010)



Classification of Disorders in Arabs (WHO ICD-10, 2010)

NBS programs in the region

- Slow progress for development and implementation of NBS programs in the region **due to cultural, legal, financial and political issues**

- In most countries in the region there is **only selective screening programs** for metabolic disorders.

- Most of the NBS laboratories in the region do **not have a complete NBS solution**

Retrospective Study in Bahrain

- For the countries where there are **no mandatory newborn screening programs**, **selective screening could be an important diagnostic tool** for diagnosis of inborn errors of metabolism
- Suspected neonates for metabolic disorders from regional hospitals are routinely referred to the Metabolic Biochemical Genetic Unit of Princess Al-Jawhara Centre in Bahrain

Retrospective Study in Bahrain

- Retrospective data examined for period **of 3 years (2008–2010)**
- The incidence of **inborn errors of amino acids, organic acids and fatty acids metabolism** in newborns **(aged 3-90 days)** suspected with metabolic disorders were investigated.
- A **total of 1645** of infants were referred and investigated for inborn errors of metabolism
- Whole blood spot were obtained by heel prick from children and spotted on Guthrie filter cards

Diagnostic Tests

- Initial and repeat MS/MS blood spot analysis
- Analysis of urinary organic acids using GC-MS
- Analysis of urinary and plasma amino acids using HPLC
- Analysis of plasma and urinary acylcarnitines using LC-MS/MS

Table 1: Classification and biochemical confirmation

Disorder	Initial and repeated LC-MS/MS screening	Biochemical confirmation
Mable Syrup Urine Disease	High valine, isoluecine and leucine	High plasma and urine valine, isoleucine and leucine
Argininemia	High Arginine	High plasma arginine
Phenylketonuria	High phenylalanine	High plasma phenylalanine
Arginosuccinic Aciduria	High arginosuccinic acid	High plasma argininosuccinic acid
Isovaleric Acidemia	High C4	High urinary isovalerylglycine
Propionic Acidemia	High C3	High urinary propionic acid
Methylmalonic Acidemia	High C3	High urinary methylmalonic acid
Glutaric Aciduria type II	High C4; C5	High isovalerylglycine

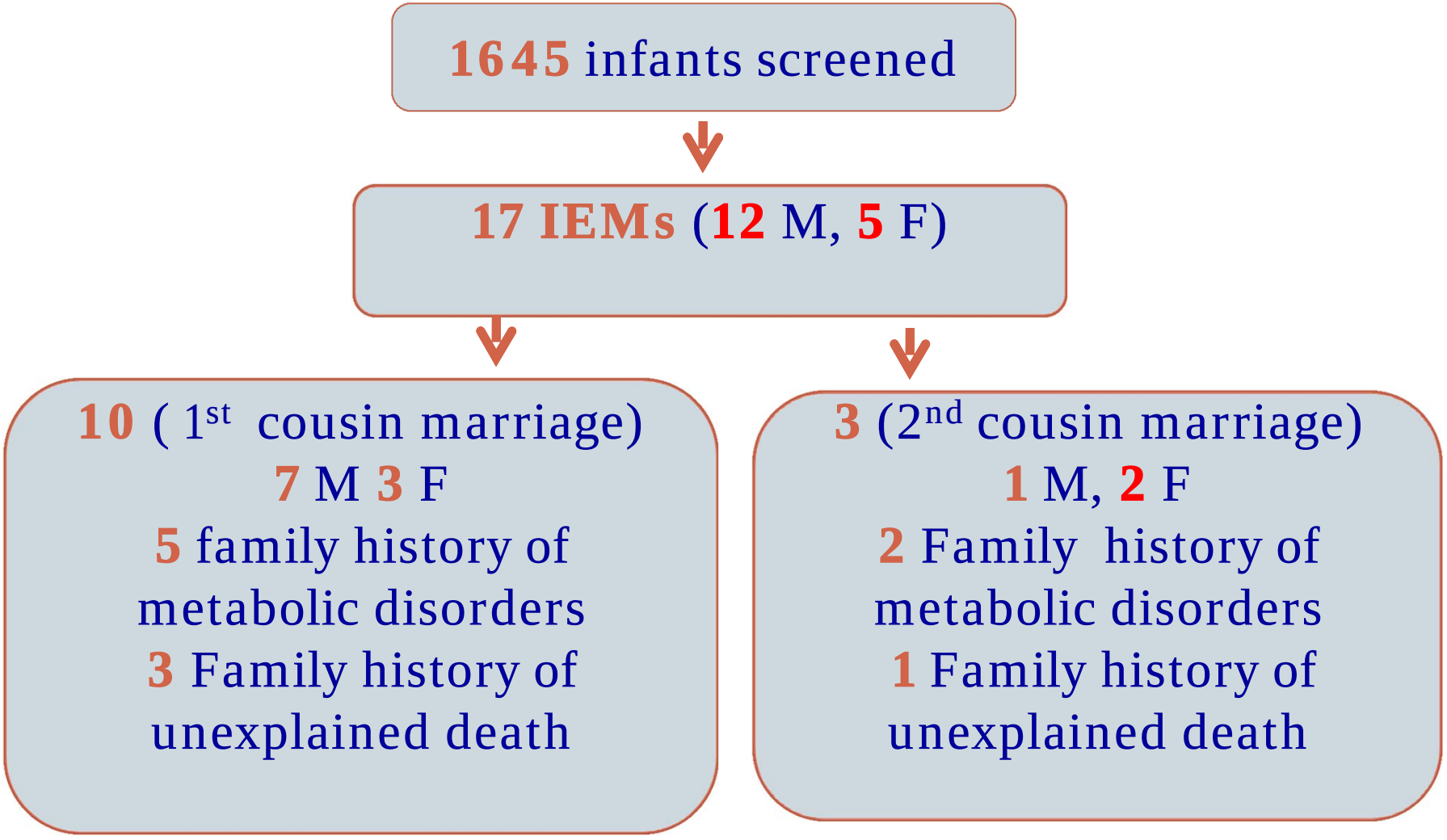
Table 2. Frequency and detection rates of amino acid, fatty acid and organic acid disorders. (51,924 live births, 17 cases, 2008-2010)

Disorder	No of Cases (Gender)	Detection rate	Incidence (No./live birth)	Incidence worldwide
Mable Syrup Urine Disease	3 (M), 1 (F)	1:548	1:12,981	1: 185,000
Phenylketonuria	1 (M)	1:1645	1:51,924	1:10,000
Argininemia	1 (M)	1:1645	1: 51,924	1:350,000
Arginosuccinic Aciduria	1 (M)	1:1645	1: 51,924	1:70,000
Isovaleric Acidemia	2 (M)	1:823	1:25,962	1:250,000
Propionic Acidemia	1 (M), 1(F)	1:823	1: 25,962	1:100,000
Methylmalonic Acidemia	2 (M), 1(F)	1:548	1:17,308	1: 100,000
Glutaric Aciduria type II	1 (M), 2 (F)	1:548	1:17,308	1:40,000

Table 3. Incidence of metabolic disorders according to the type of disorder and birth cohort.

Year	Live Birth	Amino Acid Disorders	Organic Acid Disorders	Fatty acid Disorders	Overall
2008	17,841	2 (1:8,920)	2 (1:8,920)	1 (1:17,841)	**5 (1:3,568)**
2009	17,022	2 (1:8,511)	2 (1:8,511)	1 (1:17,022)	**5 (1:3,404)**
2010	17,062	3 (1:5,687)	3 (1:5,678)	1 (1:17,062)	**7 (1:2,437)**

Impact of Consanguinity



This can be attributed to the lack of early detection of such disorders, as well as the lack of parental genetic counseling.

Conclusions

- These findings reflect the **significant contribution of consanguinity** in Bahrain in inherited metabolic disorders
- The data presented in this study are only the **beginnings of attempts** to **estimate the accurate incidence** of metabolic diseases in Bahraini population.
- These data can be regarded **as a guide to how we can provide diagnostic services for metabolic diseases** in the future.
- This study emphasizes the **important role of the specialized laboratories** in obtaining such data that to **be used for the recommendation of mass screening program** in a population at risk.
- Such **approach should be extended** for other metabolic disorders such as **mitochondrial disorders** in Bahrain.

Acknowledgment

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- Centers of Disease Control and Prevention (CDC, USA)

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