



Al Jawhara Centre for Molecular Medicine & Inherited Disorders
مركز الجوهرة للطب الجزيئي وعلم الوراثة والأمراض الوراثية

Newborn Screening, Quality Performance and Strategies for improvement in the Middle East

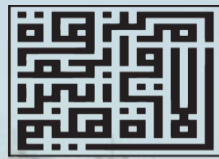
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**Associate Professor of Molecular Medicine, Managing Director of
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HH Princes Al-Jawhara Centre

College of Medicine and Medical Sciences,

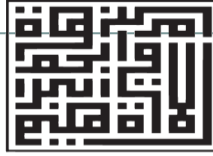
AGU- Bahrain



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HH Princes Al-Jawhara Centre for Molecular Medicine and Inherited Disorders Manama, Bahrain



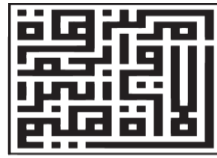
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- Consanguinity in Arabian Peninsula
- NBS Programs In The Region
- Challenges In Clinical Follow-up
- Issues With Sensitivity and Specificity
- Quality Strategies
- Research Strategies
- Further Directions



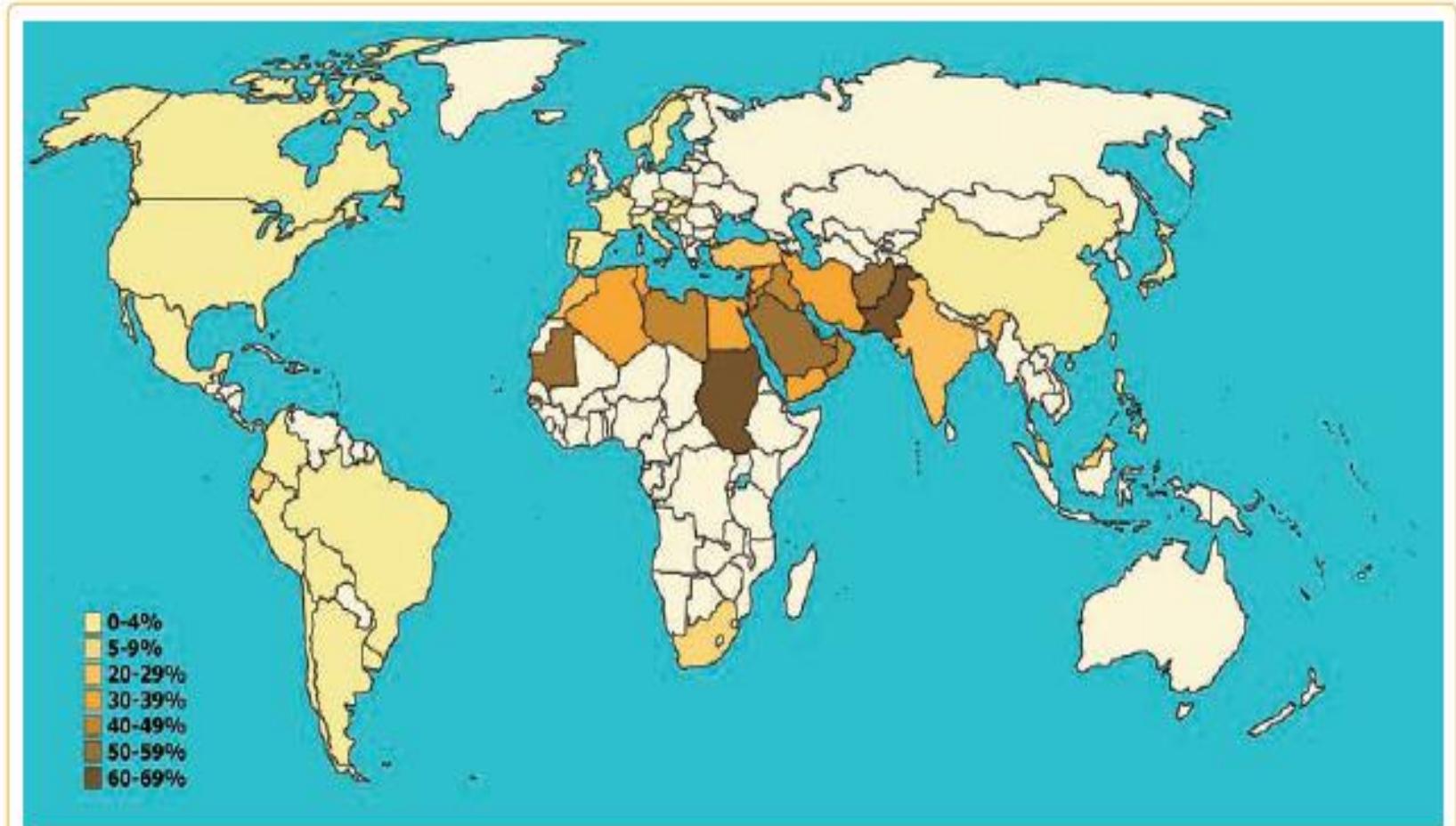
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- NBS is NOT WELL recognized as an essential, preventive public health program in the region
- It is NOT WELL appreciated that detection, diagnosis, and treatment of inherited metabolic disorders can lead to significant reductions of death, disease, and associated disabilities.
- 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**



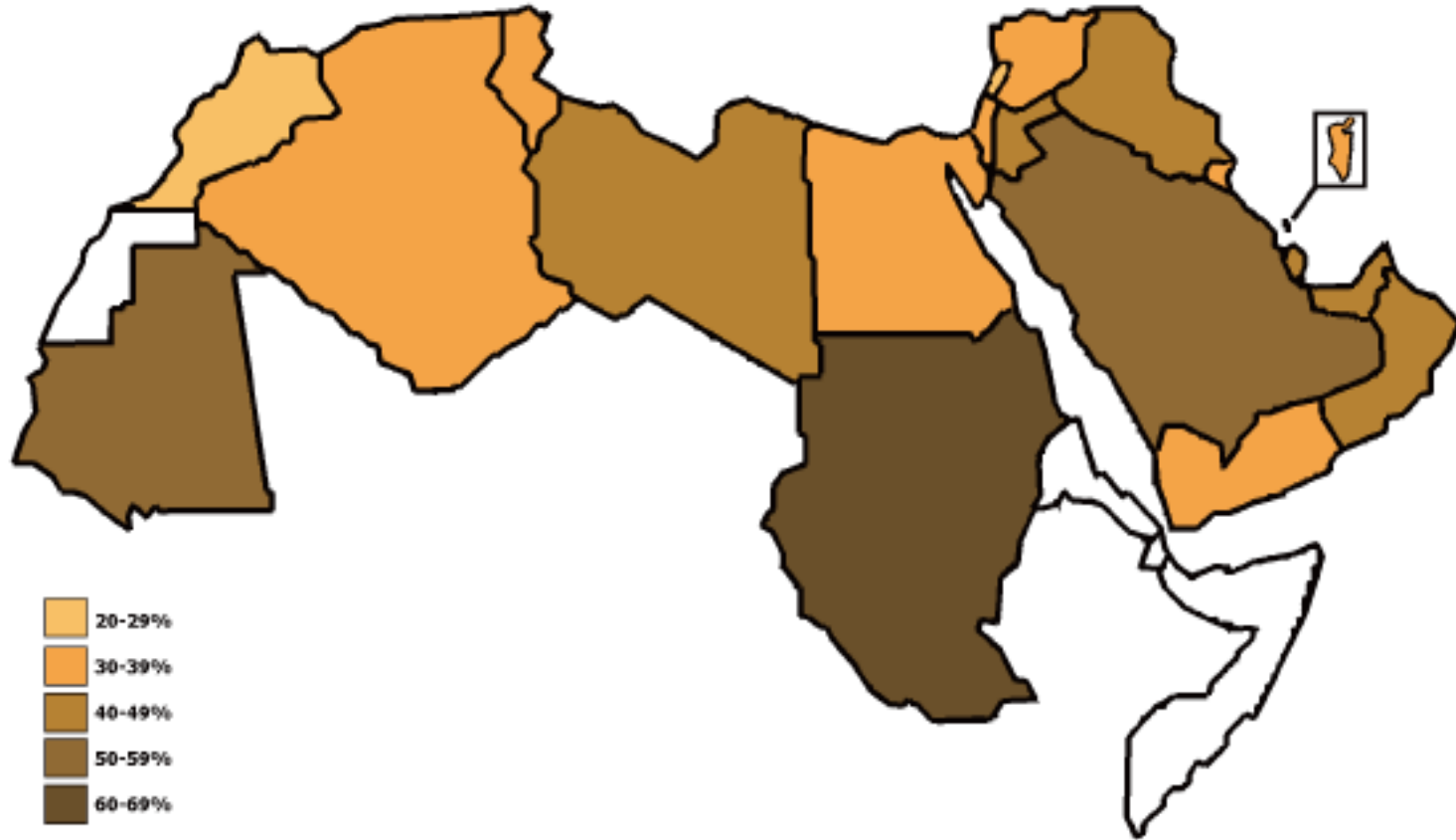
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Consanguinity

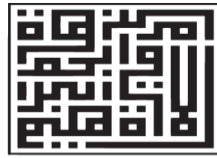




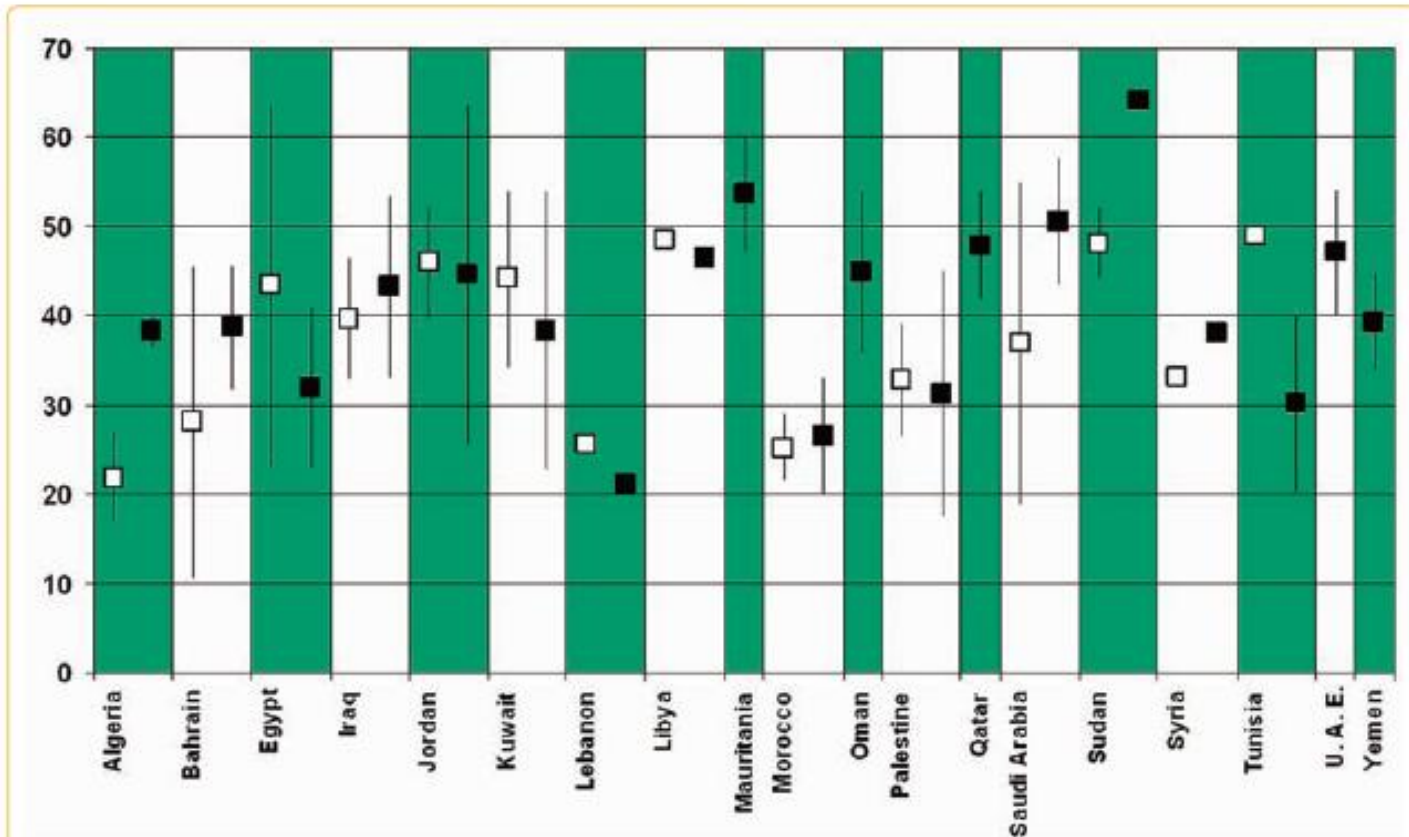
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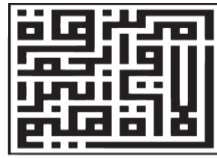
In the Arabian Peninsula, there are high percentages of consanguineous marriages and the tribal nature of marriages



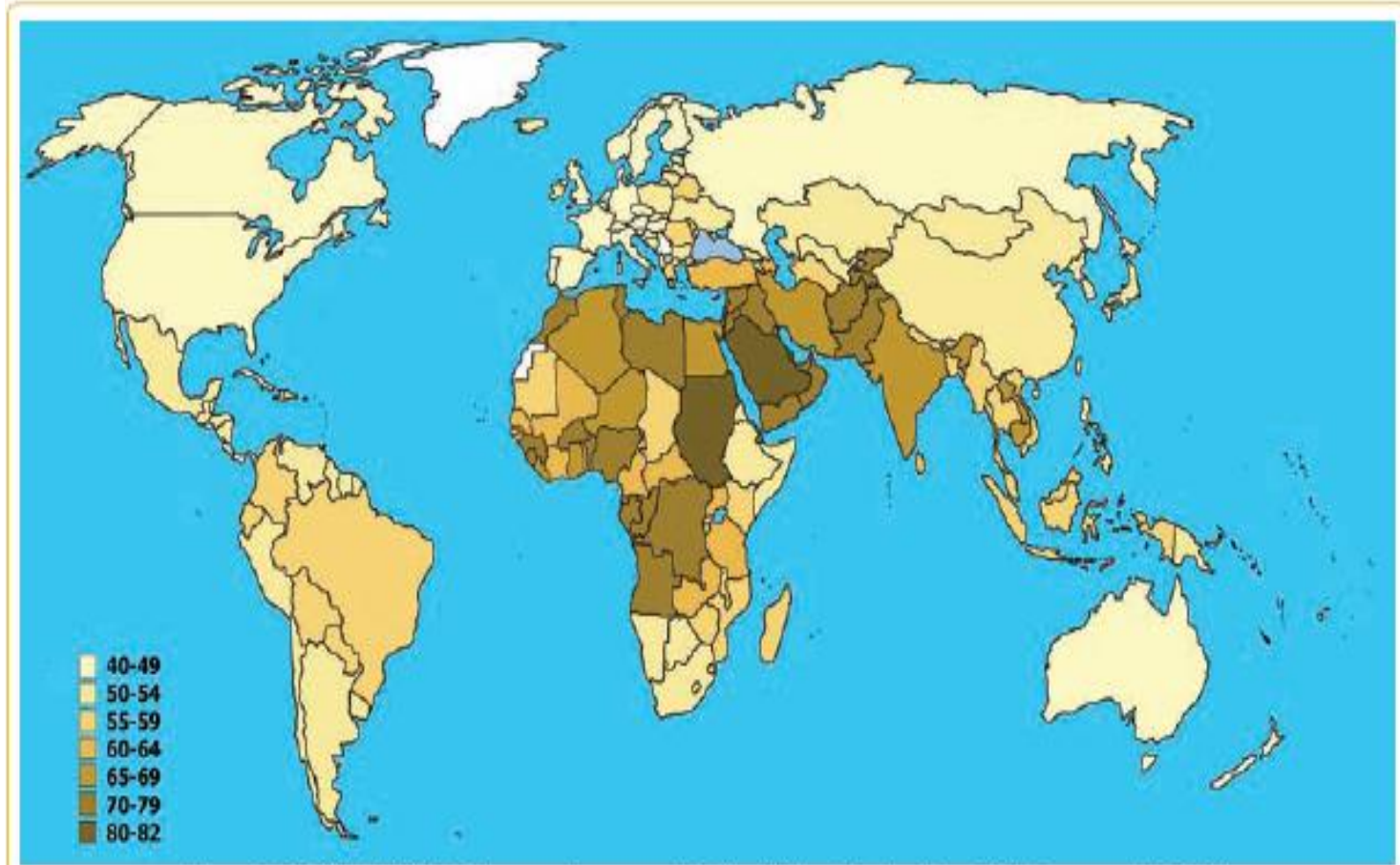
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Past (White boxes) and present (black boxes) consanguineous marriage percent in Arab States



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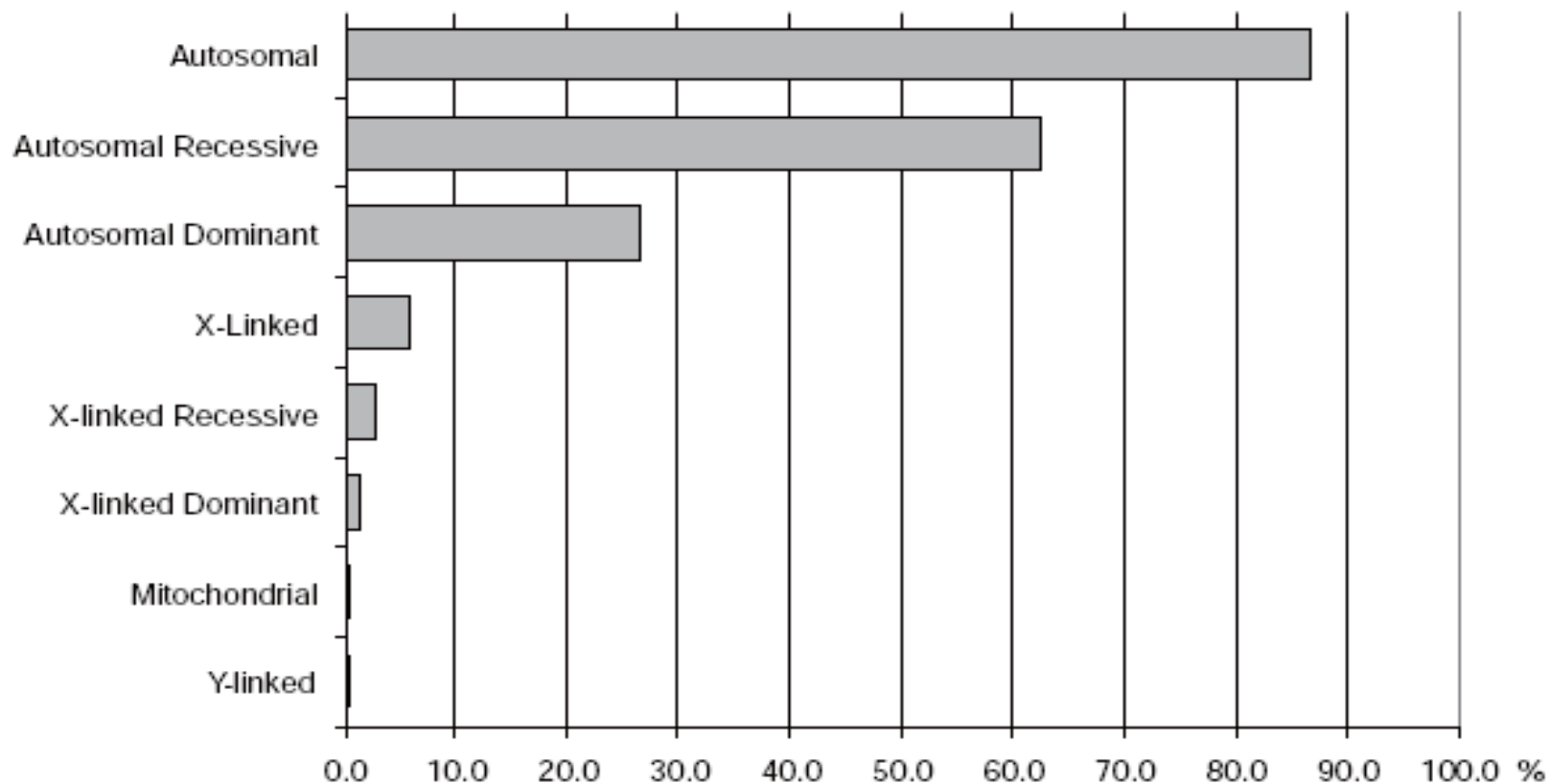


Global birth defects prevalence per 1000 live birth

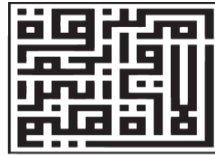


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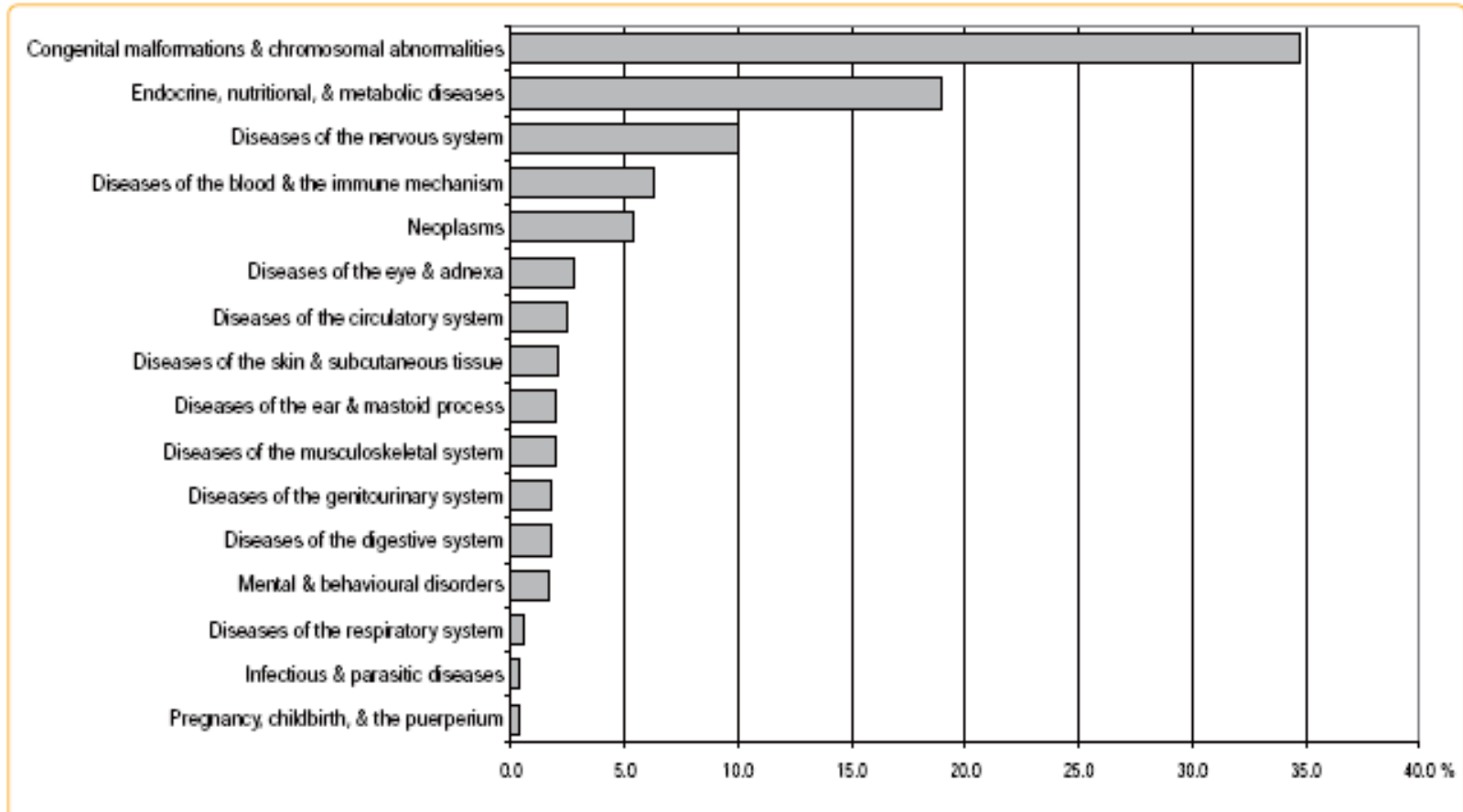
Genetic Disorders



Classification of Genetic Disorders in Arabs according to mode of inheritance (WHO ICD-10, June 2008)



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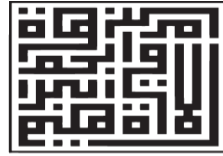
Classification of Disorders in Arabs (WHO ICD-10, June 2008)



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➤ According the statistics published by Ministry of Health in Bahrain in average there are **500 deaths among children < 5 years old** which contributes to the 22% of the total death each year.

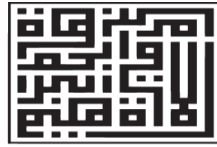
➤ The average total incidence of **post natal (3/1000), neonatal (5/1000), and infant (8/1000) mortalities is 16/1000** in the last 5 years in Bahrain (Statistics from Ministry of Health).



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5 key components of Newborn Screening

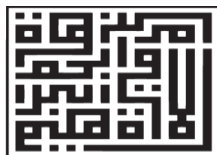
1. Screening
2. Follow-up
3. Diagnosis
4. Treatment/management
5. Evaluation



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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



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Estimated Incidence of some of Inborn errors of Metabolism in Bahrain from 1998-2007 compared to the estimated incidence in Europe and USA

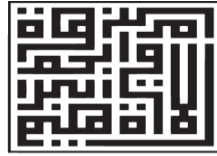
Type of the Disease		Incidence in Bahrain	Incidence in Europe and America
Amino acidopathies	PKU	1 : 10,000	1:10000
	MSUD	2 : 10,000	1:100,000
Organic acidemia	Isovaleric acidemia	2 : 10,000	1:250,000
	Propionic acidemia	1 : 10,000	1:35,000
	Methyl melonic acidemia	6 : 10,000	1:50,000
	3-methyl glutaric aciduria	2 : 10,000	1:100, 000
Mitochondrial disorder	Glyceric acidemia	6 : 100,000	1:250,000
	Fatty acid oxidation defect (CPT II)	1 : 10,000	1:75,000
	E2 defect	6 : 100,000	1:80,000
Urea cycle defect	Glutaric aciduria II	2 : 10,000	1:75,000
	Arginosuccinic aciduria	6 : 100,000	1:100, 000

Courtesy of Dr Emthesal Al-Jishi, SMC, MOH, Bahrain

Incidence of Congenital Hypothyroidism in a retrospective hospital based study in Bahrain

1:2500

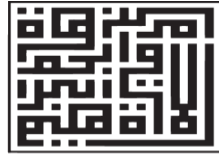
Golbahar et al. 2010 **JPEM;23:39-44**



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Challenges in clinical follow-up

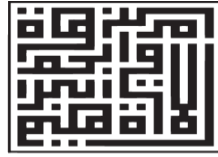
- There is no effective and timely communication among clinicians, laboratories families and public health agencies
- International and local private laboratories serving the region do not provide any means of effective follow up



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Follow up procedures

- Lab results will be immediately sent for a disorder and the results will also include:
 - ACT sheet specific to that disorder
 - FACT sheet specific to that disorder
 - List of Metabolic Specialists that are available in the country to refer to.



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Problems with Sensitivity and Specificity

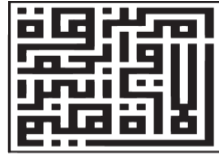
- Although in last 15 years using advanced technology in NBS the sensitivity and specificity of the newborn screening services has been immensely improved.
- However, there are limited database regarding the cut off values of in newborns in the region or for a specific country available that would lead to some false positive and false negative results



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False Positives

- Cut-off values may be too low
- TPN feedings
- Prematurity
- Sampling Error



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False Negatives

- Cut-off values may be too high
- Mild Clinical Course
- Sampling Error



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Quality Assurance Program

- Some NBS laboratory in the region still have not the best quality strategies that include quality assurance program protocols, guidelines, and standards
- NBS quality assurance program in Central of Disease and Control prevention has greatly helped the NBS laboratories and centers in the region and giving us some confidence for diagnosis of metabolic disorders
- MS/MS data project offered by Region 4 Genetics Collaborative is another source to improve the overall blood spot analysis and to set the standard to achieve the lowest achievable rates of false positive results.



CDC en Español

Environmental Health

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Newborn Screening Quality Assurance Program

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Division of Laboratory Sciences



Newborn Screening Quality Assurance Program

Log In

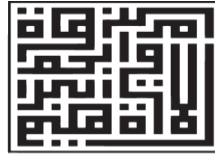
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MS/MS DATA PROJECT



Region 4 Genetics Collaborative > MS/MS Data Project



MS/MS Data Project

This laboratory quality improvement initiative strives to improve the analytical quality of newborn screening by tandem mass spectrometry (MS/MS) by:

- achieving universal tandem mass spectrometry (MS/MS) testing of newborns for a uniform panel of metabolism and congenital adrenal hyperplasia (CAH);
- improving overall analytical performance;
- setting and sustaining the lowest achievable rates of false-positive results; and
- improving and standardizing confirmatory testing and short-term follow up.

Started as a Region 4 project in 2004, this project continues to expand and currently includes states from all seven genetics regions as well as international participants from other countries.

For those interested in gaining access to the MS/MS Data Project and Laboratory Quality Improvement of NBS by MS/MS Training information, please contact David McHugh at McHugh.David@mavo.edu.

SEARCH REGION4GENETICS.ORG

GO

WHAT'S NEW

TELEMEETINGS

PARTICIPANT LOGIN

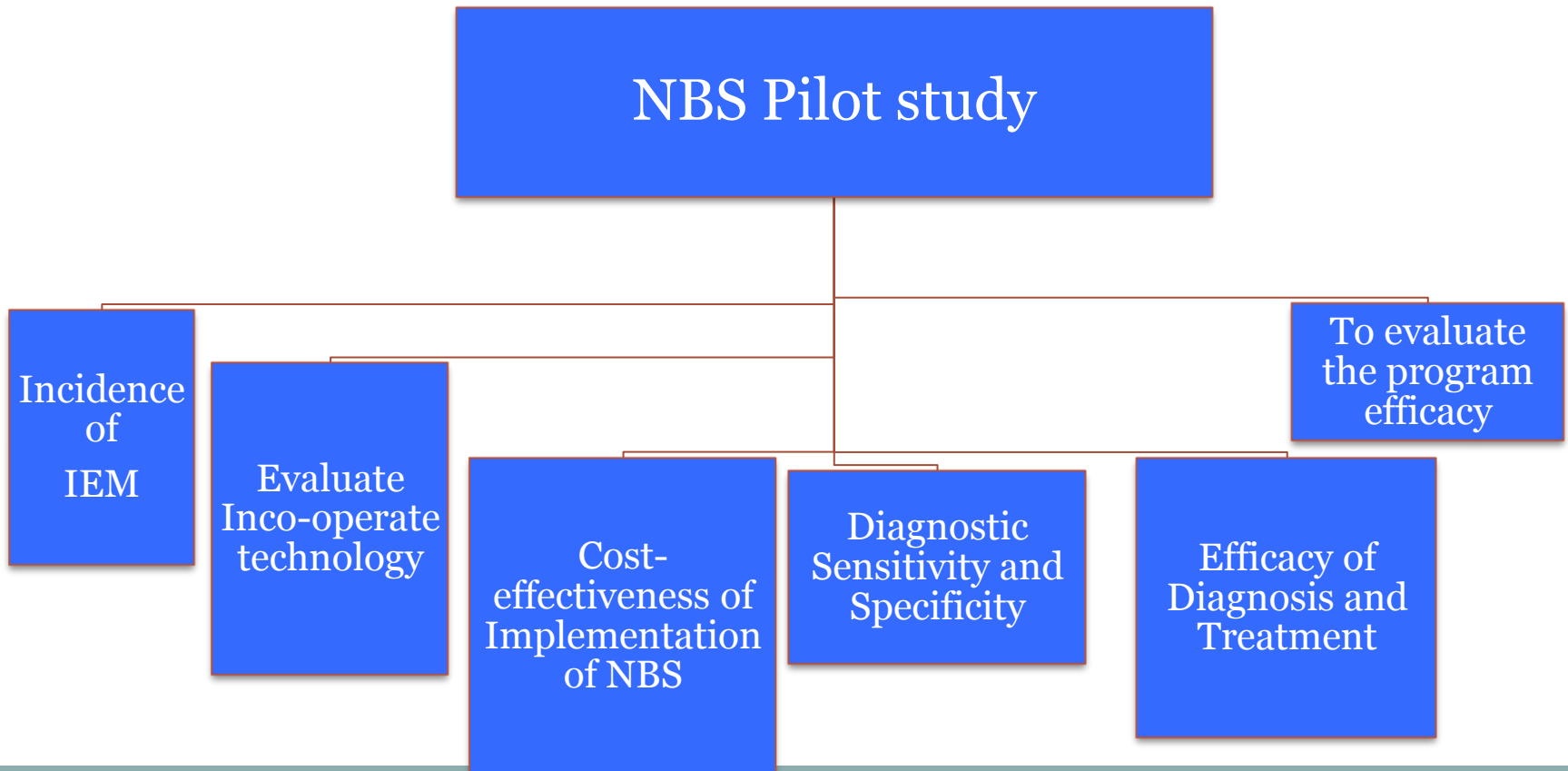
ADVISORY GROUP

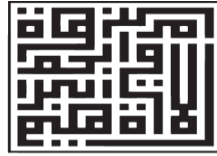
GENERAL
INFORMATION



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Research Strategy





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Further Directions

- We believe the region needs to take **massive initiatives** towards developing national strategies for NBS and using experiences of regional and international screening programs.
- There must be an initiative to create a **Central Institution for NBS in the region.**



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Further Directions

This central body could:

- Regulate the NBS program in the region
- Evaluate cultural and social issues surrounding newborn screening
- Assess those countries in the region with interest in developing and expanding newborn screening activities

Further Directions

This Central Body could also:

- Assess the capacity of the regional newborn screening by identification of areas for research infrastructure and collaboration
- Make initiatives to offer research and training programs for newborn screening in the region.
- Co-ordinate and set up strategies for some pilot studies and epidemiological assessment regarding the incidence and prevalence of those inherited metabolic and endocrine disorders within the region.
- We appreciate any help and collaboration that we can get from our colleagues in US and in Europe to go for this direction.



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Thank you for your attention

