

LESSONS FROM THE USJ NEWBORN SCREENING PROGRAM (USJ-NBS)

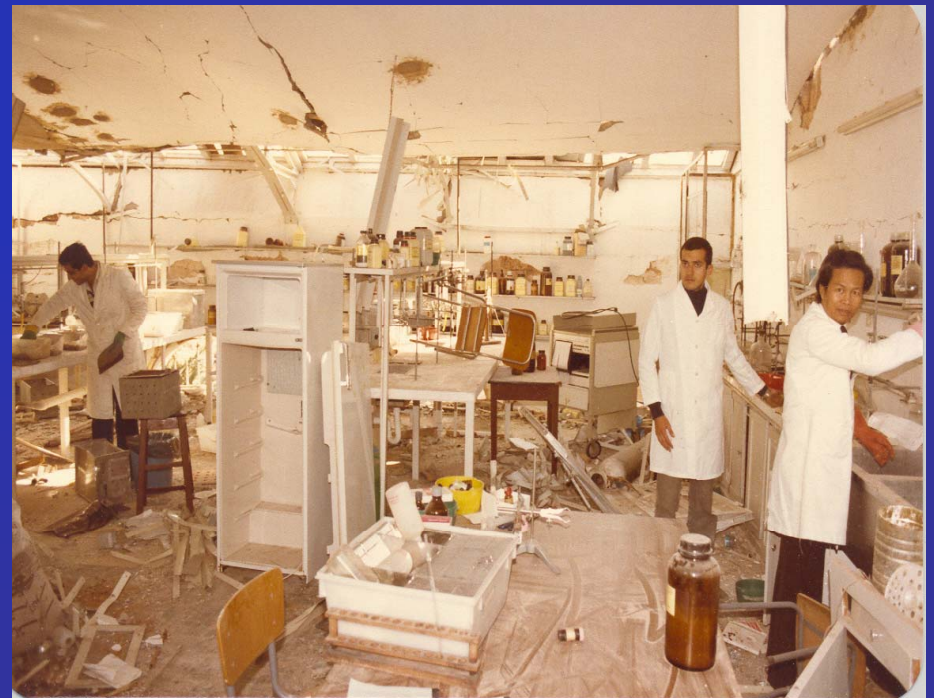
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BRIEF HISTORY OF USJ-NBS

- There are no public programs for newborn screening (NBS). About 50-60% of newborn babies never receive any screening at birth
- USJ-NBS program was initiated in 1996

Destroyed in 1975-1977





BRIEF HISTORY OF USJ-NBS

- It currently covers about 20% of all newborn babies, drawn from about 40 hospitals distributed in all regions of Lebanon
- AUBMC and other private hospitals also provide newborn screening programs for various disorders to their own users

SERVICES AND RESULTS

- In 12 years, 150,000 babies have been screened:
- Findings on first four diseases included in the NBS:
 - 92 cases of Congenital Hypothyroidism: 61/100,000
 - 830 cases of G6PD Deficiency: 1% among boys, 0.03% among girls
 - 17 cases of Phenylketonuria (1 DHPR, 13 PKU, 3 HPA): 10/100,000
 - 6 cases of Congenital Galactosemia (3 Classical, 3 Duarte): 4/100,000

RECENT EXPANSION

- Since November 2006, screening services have been extended in collaboration with European and North American centers.
- The acquisition of new technologies allows the screening of in-born errors of metabolism: fatty, amino, organic acids...
- Abnormal results are reported within 5 days. Positive results are recalled for confirmatory re-testing.
- Medical follow-up is obtained mainly at AUBMC

NEW RESULTS

Nov 2006 – Aug 2008: 22,000 newborns screened for inborn errors of metabolism

Findings:

- 38 recalls / 18 confirmed cases

PPV: 47.2%

- Cumulative incidence

82 per 100,000

NEW RESULTS

- Fatty acids defects : 3 cases
 - 1 VLCAD
 - 1 carnitine- acylcarnitine translocase deficiency
 - 1 Carnitine Update Deficiency
- Amino acid defects (except PKU): 5 cases
 - 1 Arginino-Succinic Aciduria
 - 1 Citrullinemia type I
 - 1 Homocystinuria
 - 1 Methionine Adenosyl transferase deficiency type III
 - 1 Tyrosinemia type II

NEW RESULTS

Organic acid defects : 10 cases

- 1 Propionic aciduria
- 4 Methylmalonic aciduria
- 1 Isovaleric aciduria
- 1 HMG (3-Methylglutaconyl-CoA Hydratase Deficiency)
- 1 Biotinidase deficiency
- 1 Glutaric aciduria type I
- 1 BKD (3-oxothiolase deficiency)

PRACTICAL IMPLICATIONS

- Incidence of newborn diseases relatively higher, if rare, compared to Western epidemiology: persistence of consanguinity and endogamy.
- The USJ laboratory is slowly becoming an accredited training center on recent screening technology for the MENA region.

PRACTICAL IMPLICATIONS

- Challenge: Mandating NBS for all babies in Lebanon. Meanwhile, best practice for all physicians dealing with newborn children
- Cost is still out-of-pocket, but relatively small (about 12 USD, 10 before 2006) compared to the major consequences of ignoring an early diagnosis.



THANK YOU