National Neonatal Screening Program in UAE (1995-2005)

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MOROCCO 2006

POPULATION : 3,769,080 , BIRTH RATE : 65,000

CONSANGUINITY RATE : 54% , FIRST – COUSIN : 35%

Start

The program started by screening for :

- Phenylketonuria in January 1995.
- Congenital hypothyroidism in January 1998.
- Sickle cell disease in January 2002 & March 2005.
- Congenital Adrenal Hyperplasia in January 2005 .

Protocol

The following protocol covers all stages of the screening program:

a. Invitation for screening : NS Form

b. Specimen collection : ≥ 72 hours , Heel Prick

c. Laboratory analysis : Delfia Fluorometric , HPLC
UK NEQAS

d. Follow up of positive cases : Recall System

e. Management of affected infants

f. Systematic evaluation of all phases of the program

Evaluation

A- Coverage (% uptake).

B- Timeliness of the screening program indicators (age of sampling , time of delivery of the specimen to Lab , time taken by the lab to produce the result , age of recall and age of treatment initiation) .

C- Unsatisfactory Specimen Quality.

D- Indicators for evaluation of validity of test (recall rate, the apparent sensitivity, specificity and positive predictive values).

E- Follow up results

Compared to the international standards

Results

% Uptake (Coverage):

Our results:

Target :99% of liveborn infants whose births are notified

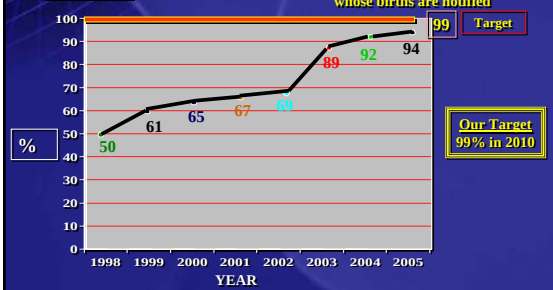

Fig 1: -% Uptake in UAE during 1998,1999, 2000 , 2001 , 2002 , 2003 , 2004 and 2005 compared to the international standards

PKU

Categories	Borderline Ph 3-4 mg/dl		Abnormal Ph >4 mg/dl		Total
	No	%	No	%	
Classical PKU	-	0.0%	26	92.9%	26
Benign Hyperph	8	12.5%	2	7.1%	10
Transient	3	4.7%	-	0.0%	3
False +ve	53	82.8%	-	0.0%	53
Total +ve screening	64	100%	28	100%	92

Table (1): Follow up of positive screened PKU cases(JAN 1995 - DEC 2005)
(n=385,135)

Congenital Hypothyroidism

Categories	Border TSH 10-25mU/L		Abnormal TSH >25mU/L		Total
	No	%	No	%	
Confirmed	11*	11.7	177	89.8	188
Transient	38	40.4	9	4.6	47
False+ve	40	42.6	4	2	44
Failure to recall	5	5.3	7	3.6	12
Total +ve Screening	94	100%	197	100%	291

*6 cases were defaulters infants whose ages are more than one month when screened.

Table (2):Follow up of positive screened Congenital Hypothyroidism cases (JAN 1998 - DEC 2005).
(n=368,993)

Sickle Cell Hb

Type	Hb Diseases		Hb Carriers	
	No Identified	Prevalence/ 100	No Identified	Prevalence/ 100
1- Sickle Hb	83*	0.06%	1154	0.9%
2- Non-Sickle Hb : Hb C	5	-	42	0.03%
Hb D	35	0.03%	487	0.38%
Hb E	1	-	10	-
3- Unidentified band	-	-	7**	-

* Sickle Cell Diseases: include 75 cases confirmed as homozygous SS and 8 cases confirmed as S/B° - thalassemia.
** 2 diagnosed as Hb O Arab by family study

Table (3) : Prevalence of confirmed sickle cell diseases, non-sickle hemoglobinopathies and hemoglobinopathy carriers during JAN 2002- DEC 2005
(n=128,148) .

Congenital Adrenal Hyperplasia

	Term	Preterm	All newborn
Screened population (n)	9,421	569	10,000
Recalls (n)	9	16	25
Recall rate	0.1%	2.8%	0.25%
CAH confirmed (n)	3	1	4
False positive recalls (n)	6	15	21
Positive predictive value	33%	6%	16%

Table (4):Follow up of positive screened Congenital Adrenal Hyperplasia (JAN 2005 - DEC 2005).

Follow up results :

Since the implementation of the program (from January 1995 until DEC 2005) : 385,135 infants were screened with relative incidence of :

- 1:1963 for *congenital hypothyroidism* (188 cases were prevented from mental retardation).

- 1:14,812 for *classical PKU* (26 cases were prevented from mental retardation).

- 0.06% for *sickle cell disease* (83 cases) and 0.9% for *sickle cell trait* (n=1154) in our program .

* Pilot study of *congenital adrenal hyperplasia* : *4 cases (3 males & 1 female) with the classic type .*

Future Plans

* Increase the uptake rate to reach 99 % by the year 2010 .

*Additional of others tests to the program:.

- Maple syrup urine Disease (MSUD)
- Galactosemia
- Tandem Mass Spectrometry
- Hearing NS


Thank you