

Abstracts of Bahrain Based Research papers presented in the International Conferences

1.

Newborn Screening by Using Mass Spectrometry.

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Introduction: Several newborn screening programs exist on wide scale in different countries. These programs vary widely, detecting disorders before frank symptoms of disease exist. Screening programs are also helpful to determine the real frequency of the diseases in population.

Material and Method: We applied newborn screening to 1000 Bahraini newborns taken at random for two years. The blood samples were collected from infants' heel on the third to fifth day after birth, on special card and send to the laboratory in King Faisal specialist hospital in Saudi Arabia, where Electrospray Tandem mass spectrometry (ESI-MS/MS) Ms/MS was applied. It analyses blood samples to thirty metabolic diseases such as amino acids, organic acids, and carnitine esters etc. Abnormal results were received in 48 hours to start treatment.

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Results: We analysed 1000 Bahraini newborn, out of which 21 infants were found to have abnormal results. Ten infants were definitely sick. In five cases the follow up samples showed normal results, and those infants are normal until now. Six cases were lost for follow up. The preliminary results showed ten abnormal cases out of 1000 newborns, which give the incidence of 1%. We expect to have 100 sick infants with metabolic diseases out of the 1000 newborns annually. Diseases such as maple syrup urine disease, medium chain acyle co, a dehydrogenase deficiency, primary carnitine deficiency, methyl malonic acidemia, methylene tetrahydrofolate reductase deficiency were found to be common. These results are not comparable with other studies as we are studying large number of diseases at neonatal period, each of these diseases have low frequency rate.

Conclusion: Prevalence rate of metabolic diseases is high among our newborn, and a metabolic screening program is essential to provide better and cost effective medical care for patients. The clinical picture of the diseased infants will be presented.

2.

Premarital Counseling, An Experience from Bahrain

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Introduction: Bahrain was the first country in the Arabian Gulf to recognize the burden of blood genetic disorders. The studies shows that 1-2% of newborn have sickle cell disease and 11-18% are carrier, while the carrier rate for the beta thalasaemia is 2%. In an attempt to reduce the incidence of babies born with these diseases, the genetic clinic started the service of Premarital screening and counseling in 1985. In 1992 the Ministry of Health expanded this service to cover all the primary health care centers and identified couples at high risks were counseled and advised.

Material and Methods: The services were preceded by an information and training course to all doctors and nurses in health centres. This was followed by mass media campaign on the availability of such services. A special risk assessment sheet was designed, which included information about sex, age, education, occupation, consanguinity, medical and surgical history, infection history, sexually transmitted disease, family genetic history, habits as

smoking, alcohol are asked for. This was followed by general physical examination. The investigations include full blood count, blood groups, haemoglobin electrophoresis, glucose 6 phosphate dehydrogenase deficiency, rubella AB, some cases are screened for venereal disease hepatitis and Aids. On the second visit the results are discussed with the client and counseling and management are given.

Results: By studying and analyzing 500 formats for 500 clients taken at random: Male/Female ratio 52.5/47.5 the mean age at marriage for male is 26.5 year and for female 21.9 years, minimum age is 15 years and maximum is 45. Regarding Consanguinity: 23.2 were first cousins 1.5% were second cousins and 3% far relatives. Regarding blood diseases SCD was found in 1.6%, SCT - 13%, Beta thal trait 2%. Average A2 in Beta thal trait is 5.6%. G6PD deficiency was found in 26% of samples, 8% of attended couples were found to be at risk of having affected offspring. The consanguinity rate among them was 15%.

Conclusion: All clients benefited from premarital counseling session especially the couples at risk.

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