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Genetic screening and diagnostics with special emphasis on hemoglobinopathies

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The past couple of decades have witnessed enormous efforts in medical genetics which improves the health and quality of life of individuals, their families, communities and societies in general. These efforts were crowned by the information obtained from mapping the Human Genome. Genetic screening for hereditary diseases is having a great impact on the health and well being of the populations in general. Genetic disorders due to chromosomal abnormalities as in Down syndrome; due to single gene mutations as in sickle cell anemia and others like cancers are seen. Screening programs that cover newborn screening, prenatal diagnosis and carrier testing are done to detect chromosomal abnormalities, DNA changes and protein/biochemical changes. In the USA, programs are USA endocrine disputor program, national breast and cervical cancer early detection program, HIV in US emergency departments and newborn metabolic screening programs. In Saudi Arabia, the prominent programs are the newborn screening, premarital screening for sickle cell anemia, thalasemia and breast cancer screening. Other countries perform similar programs according to their needs. Ethical issues raised in these screening programs include privacy, misuse of genetic information, testing children for a carrier state and prenatal diagnosis especially for non-disease traits or sex. The recent advances have paved the way towards gene therapy and personalized medicine. In this presentation, an overview of genetic screening and diagnosis with special emphasis on genetic screening and diagnosis of hemoglobinopathies is given.

Biography

Gwiria M H Satti completed her PhD from Leeds University, Biochemistry, 1987. She was teaching Biochemistry to medical students for 36 years at University of Khartoum, King Faisal University, Dammam-Saudi Arabia and at KFMC, King Saud bin-Abdulaziz-University for Health Sciences, Riyadh-Saudi Arabia. She was a PI for a Malaria project in the Sudan between 1991 and 1998 in collaboration with staff from University of Copenhagen and ICAPB, Edinburgh. Seventeen publications came out from that work. She is currently involved in a potential anti-diabetic plant research at KFMC and medical education research..

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