## **Down Syndrome in Oman**

We present, here, the first relevant study of Down syndrome (DS) in Oman between 2000 to 2004. The study was consisted of four parts: 1. Epidemiological survey: to determine the birth prevalence of DS in the population. The data were based on Cytogenetic Registry provided by the Genetic Unit in the Ministry of Health. 2. Cytogenetic study: the aim was to determine the different types of cytogenetic abnormalities in DS children. In addition, birth prevalence of DS in the population was documented using the cytogenetic data as a national registry for DS in the Omani population. 3. Molecular genetic study: Using high polymorphic STR markers the parental and meiotic origin of nondisjunction and the frequency and localization of the crossovers were analyzed. DNA samples were obtained from buccal smears of the DS children and both parents at the time of completing the questionnaire. 4. A case control study: this is based on the cases with free trisomy 21 identified by the Cytogenetic Laboratory through all regional hospitals in Oman. The controls were recruited as matched for the delivery of an unaffected child in the same year and in the same Health Institute, Governorate or Region.

Dr. Salma Al Harrasi is a genetic Laboratory Consultant at the National Genetic Center, Oman. She holds PhD in Human Genetics and by 2013 she was appointed as Head of Genetic Laboratory, with the main goal to ensure the provision of high quality genetic laboratory testing to patients and families affected by genetic diseases in Oman.



978-3-659-84223-8

