

Down syndrome is the most common Aneuploidy in humans which is associated with developmental delay, mental retardation and several characteristic physical features. This study is aimed to determine the proportion and types of chromosomal abnormalities in patients with Down syndrome in Duhok province, Iraq, and to study the clinical profile of these patients. A retrospective analysis was performed on the case records of 86 patients confirmed clinically as Down syndrome from October 2014 to April 2015, for all enrolled cases cytogenetic analysis had done at Research Centre at College of Medicine\ University of Duhok. Among the 86 cases of Down syndrome presented over a period of 6 months, non-disjunction was present in 79 (91.9%) cases, translocation in 5 (5.8%) cases and Mosaicism in 2 (2.3%) cases. The age of the enrolled patients were ranged from 1 day to 35 years, from these patients, 37 (43%) were males and 49 (57%) were females with male to female ratio of 1:1.3. The maternal ages at the time of delivery of index babies were ranged from 21 years to 47 years. The most prominent characteristic features noted were epicanthic folds (80.2%), upslanting palpebral fissures (70.9%), protruding tongue (67.4%), sandal gap (64%), depressed nasal bridge (62.8%), and flat facial features (58.1%). Congenital heart disease seen in (26.7%) and hypothyroidism seen in (10.5%)

Down syndrome mostly result from non-disjunction and efforts to establish early diagnosis and a proper screening for high association with systemic anomalies should be undertaken among the Down syndrome patients in our population.