Estimation of Common Chromosomal Aneuploidies in the Population of Bahrain: 20 Years of Findings in the Genetic Laboratory

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Objective: To evaluate the incidence of common chromosomal aneuploidies in Bahrain during the last 20 years (1999-2019).

Design: A Retrospective and Descriptive Study.

Setting: Genetic Laboratory (now National Genome Center) at Salmaniya Medical Complex, Bahrain.

Method: A total of 5,153 patients' karyotypes and clinical reports were reviewed retrospectively. Conventional cytogenetic analysis was accomplished for karyotype reporting at the 400-band resolution level by using Giemsa staining technique. Fluorescence in situ hybridization (FISH) assay was used as well on selected cases.

Results: Out of 5,153 patients we uncovered 758 cases with various structural and numerical abnormalities. Numerical abnormalities (aneuploidies) were uncovered in 529 (69.7%) subjects. This includes 476 (62.8%) cases with autosome aneuploidies and 53 (6.9%) cases with sex-chromosome aneuploidies. Trisomy 21 (Down's syndrome) is the most common autosomal aneuploidy at 404 (53.3%) with an estimated population incidence rate of 1:701 live births. Trisomy 18 (Edward's syndrome) found in 53 (6.9%) patients, and trisomy 13 (Patau's syndrome) at 4% (19 patients) of autosomal aneuploidies. Incidence rate indicates 1:5343 for trisomy 18 and 1:16660 for trisomy 13. In contrast, our findings for the sex-chromosome aneuploidies includes 17 (2.2%) patients with Klinefelter's syndrome (47, XXY) and 28 (3.7%) patients with either standard Turner's syndrome (45, XO), 9 (1.2%), or other various mosaic Turner's syndrome, 19 (2.5%). Estimated incidence rate for Turner's syndrome is 1:10115 and for Klinefelter's syndrome is 1:16660. Finally, we uncovered 8 (1.1%) patients with rare sex chromosome aneuploidies or combined sex chromosome and autosome aneuploidies with an estimated overall incidence of 1:35402.

Conclusion: The most common autosomal aneuploidies in Bahrain is attributed to trisomy 21 (Down's syndrome) followed, with significantly less frequency, by trisomy 18 and 13. For sex-chromosome aneuploidies, various types of Turner's syndrome (including mosaics) are the most common followed by the Klinefelter's syndrome.

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