High frequency of Klinefelter syndrome in a cohort of Sri Lankan males with azoospermia and oligozoospermia

Samaranayake UMJE[1]*, Sirisena ND [1,2], Malalasekera AP[1], Dissanayake VHW[1,2,3]

[1]*Department of Anatomy, Faculty of Medicine, University of Colombo.*
[2]*Human Genetics Unit, Faculty of Medicine, University of Colombo.*
[3]*Asiri Centre for Genomic and Regenerative Medicine, Asiri Surgical Hospital, Colombo.*
**eshana_sam@live.com.sg*

Klinefelter syndrome (KS) is the most common sex chromosomal aneuploidy which affects 1:667 new born males. It is a major cause of male factor infertility due to the associated testicular atrophy. This study aims to describe the karyotypes identified in a cohort of Sri Lankan infertile males referred for cytogenetic testing prior to undergoing testicular biopsy. The karyotype reports of males with severe oligozoospermia (sperm cell count<5×106 cells/ml in seminal fluid analysis) or azoospermia, who were referred for karyotyping between January 2010 and February 2019 were maintained prospectively in an anonymized database and analysed retrospectively. All patients were referred from a single urological practice prior to undergoing testicular biopsy. Karyotyping was performed on routinely cultured lymphocytes after GTG-banding according to the guidelines of the International System for Human Cytogenetic Nomenclature (2016). A total of 69 infertile males underwent karyotyping. Among them, forty (58.0%) had azoospermia and 29 (42.0%) had severe oligozoospermia. Abnormal karyotypes were seen in 14 (20.3%) males, comprising of 11 (78.6%) with KS (47, XXY) and 3 (21.4%) with 46, XX karyotype. Polymerase chain reaction for SRY gene was positive in one male with 46, XX karyotype. Among the KS patients, 6(54.6%) had azoospermia and 5 (45.5%) had severe oligozoospermia. KS accounted for more than two-thirds of the chromosomal anomalies in this cohort of infertile males. Cytogenetic testing of severe oligozoospermic and azoospermic males may help avoid extensive and invasive investigations in both partners. Multicentre studies with a larger sample size would provide more generalizable data on the karyotype profile among Sri Lankan infertile males.