

The prevalence of the Klinefelter syndrome and its variants in a specific Hungarian infertile male group

Anthropologiai Közlemények 47 (2006), 51–55.

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Abstract: Data from the Hungarian Central Statistical Office (KSH) show that infertility affects about 150.000 couples in Hungary. The reports have shown that female and male contribution to the problem is equal. Chromosomal abnormalities are one of the major causes of male infertility. The most common abnormality is the Klinefelter syndrome. The aim of this study was to estimate the prevalence of Klinefelter syndrome in patients with azoospermia in a specific sample. Between 2003–2006 we investigated sex chromosomal abnormalities in 62 infertile men that came from urology clinics or were investigated in paternity and crime cases. Karyotyping was performed on lymphocyte preparations of men with diagnosis of infertility. The traditional cytogenetic methods (QFQ-, GTG-, CBG-band) were combined with fluorescence in situ hybridisation. Among the investigated subjects we found three 47,XXY, one 47,XXY/49,XXXXY, and two 48,XXYY syndromes. There are few reports on very low rate spermatogenesis in occasional patients with Klinefelter syndrome, but all of our patients were azoospermic.

Keywords: Sex chromosome abnormalities, Klinefelter syndrome, Infertility, Azoospermia.