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Elective Termination Decision in Sex Chromosomal Abnormalities-Current Situation in Decision Making Process

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Sex Chromosome Abnormalities (SCAs) are the most common genetic disorder with a frequency of 1/400 or 1/500 live births [1]. Reproductive decisions based on genetic counseling of SCAs are complicated. Prenatally diagnosed diseases with uncertain consequences or moderate severity is the main subject of this complex decision making process. What is the most recent scientific approach in the elective termination of SCAs?

Reproductive decisions post genetic counseling, in all genetic conditions; represent a dynamic interaction between patients, obstetricians and genetic counselors. A limited number of principles, guidelines and standards must be applied when counseling in regard to "testing for fetal genetic disease." These principles dictate that genetic counseling should be non-directive and unbiased and that parental decisions should be supported regardless of the reproductive choice [2].

Elective termination rates subsequent to prenatal diagnosis differed greatly depending on the sex chromosome abnormality. The primary offender was 45, X (54%), followed by 47, XXY (46%), 47, XYY (29%), and 47, XXX (17%) in a local study [3]. In our recent study, a total of 60 pregnancies (0.80%) with SCA were evaluated. Turner syndrome was the most commonly diagnosed SCA in prenatal diagnosis (60%). The most common reason for referral in relation to pregnancies with Turner syndrome was cystic hygroma observed via ultrasonography. Of 14 pregnancies presenting with a prenatal diagnosis of SCA (Turner syndrome: 7, Klinefelter syndrome: 5, Mosaic Turner syndrome: 2), 12 (85.7%) were terminated. The ratio of SCA in the prenatally diagnosed cases was similar to that reported in the literature. Although the ratio of terminated pregnancies with Turner syndrome was similar to that reported in European countries, all the pregnancies with Klinefelter syndrome chose termination, which showed a regional difference in Turkey [1].

Interestingly, recent studies have revealed a specific, socially functioning, profile of males with SCA with the suggestion being that it presents vulnerability towards autism [4]. Agreements for termination of pregnancy for sex chromosome or moderate handicap anomalies, when the law only permitted termination of pregnancy for serious and incurable fetal anomalies, were motivated by perceived parental suffering, which was considered the priority. These handicaps were not reversible and could quite conceivably have led to more serious incurable disorders. Other studies have also found that decisions for termination of pregnancy in cases of mild and minor congenital abnormalities have often raised major ethical dilemmas and have not always been in accordance with the regional laws [5,6].

Consequently, an elective termination decision, following prenatally diagnosed SCAs and genetic counseling, is still a hotly debated topic which requires tight and insightful collaboration from all parties concerned as well as updated and relevant genetic information.

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