The Meaning of Early Knowledge of a Child's Infertility in Families with 47,XXY and 45,X Children

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ABSTRACT: Nine girls with X or partial X monosomy and fourteen boys with 47,XXY karyotypes were identified through the consecutive chromosome screening of 40,000 newborns over a ten-year period. The probable infertility of these children emerged as a central concern in annual psychiatric interviews with parents. Parental grief over the anticipated loss of this choice for their child, whether or not openly avowed within the families, influences the development of self-image and self-esteem in the child and, if not appropriately confronted, blocks the acceptance process necessary in the child as preparation for parenthood by means of adoption.

This article identifies and describes a group of parents having early knowledge of their child's anticipated infertility. Study of this group advances understanding of how this knowledge is related to family and individual development. How, and at what developmental stages this knowledge is integrated may have lifetime effects, since assumption of fertility is part of an individual's body image and self-esteem, and since the grieving and acceptance of one's infertility is known to be a necessary step to successful adoption. These parents were identified while conducting a longitudinal sex chromosome aneuploidy study started in 1964. The purpose of the study was to define the prognosis of individuals with sex chromosome abnormalities (47,XXY, 47,XYY, 47,XXX, 45,X and mosaics) in an unselected population. Most of our knowledge, until then, was limited to findings on a selected population that had come to the notice of the medical profession because of...
problems. These findings have been known to most of us as "syndromes"—the Klinefelter syndrome (47,XXY), Turner syndrome (45,X), Triplo-X syndrome, and XYY syndrome. Forty thousand consecutive newborns were screened for sex (X) chromatin; the last 15,641 were also screened for Y-chromatin. Discrepant cases were confirmed by chromosome analysis. Between 1964 and 1974, 68 infants with sex chromosome anomalies were identified. Seven died in the neonatal period, and of the remaining 61 newborns, the parents of 51 consented to participate in a prospective family development study.

Initially, with the attending physician's permission, a conference was held with the parents. The parents were told that a chromosomal variation had been found, and that this finding might be of importance for a proportion of the children with the variation, although its significance could not be predicted. All parents were reassured that mental retardation was not to be expected, as it is in Down Syndrome. The parents of 47,XXY boys and of 45,X and partial X monosomy girls were usually informed of the probability that their child would be infertile. The parents who joined the study were assured that, should developmental problems occur, they would be discussed and all possible remedial steps taken.

The children were seen quarterly in the first year, semi-annually in the second and third years and annually thereafter. Observations included physical and psychological examinations. In addition, home visits with the family members permitted close contact with their environment. School records were examined, and school visits were made as indicated.

The project is called a "Family Development Study," as parents and siblings are included whenever possible. Siblings have provided a measure of control for factors (both environmental and genetic) other than sex chromosome abnormality.

The evaluating staff originally consisted of a pediatrician, a social worker, a clinical psychologist, and a speech pathologist. Since 1974 a child psychiatrist has been part of the team. Over the past eight years a clinical psychiatric interview has been part of each family's visit, and psychiatric consultation has been available to each family in the study according to its need. The information gathered during these interviews has had particular relevance to the impact of the study on participating families as experienced at different stages in the identified child's development. At a weekly team conference, the psychiatric aspects of each family's development were reviewed. This enables the team to remain sensitive to the psychological impact that