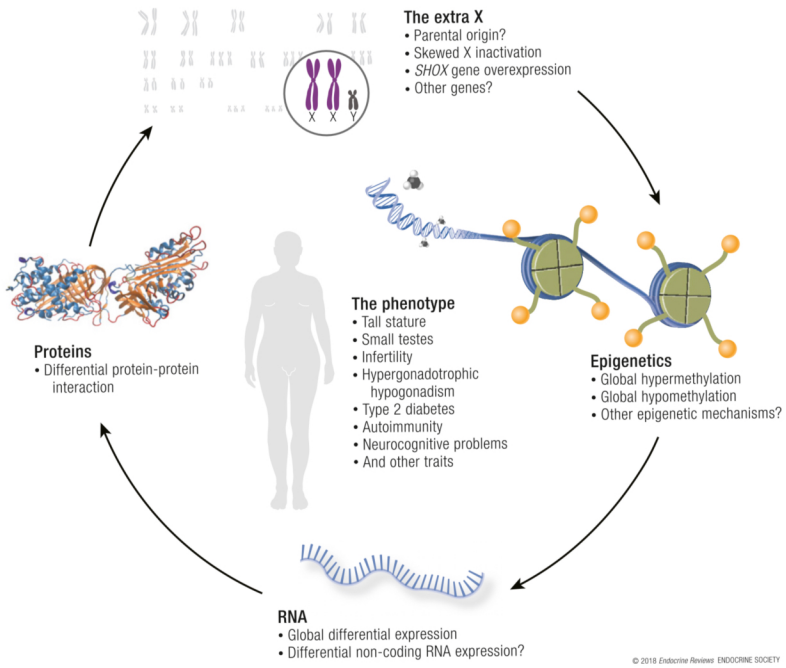
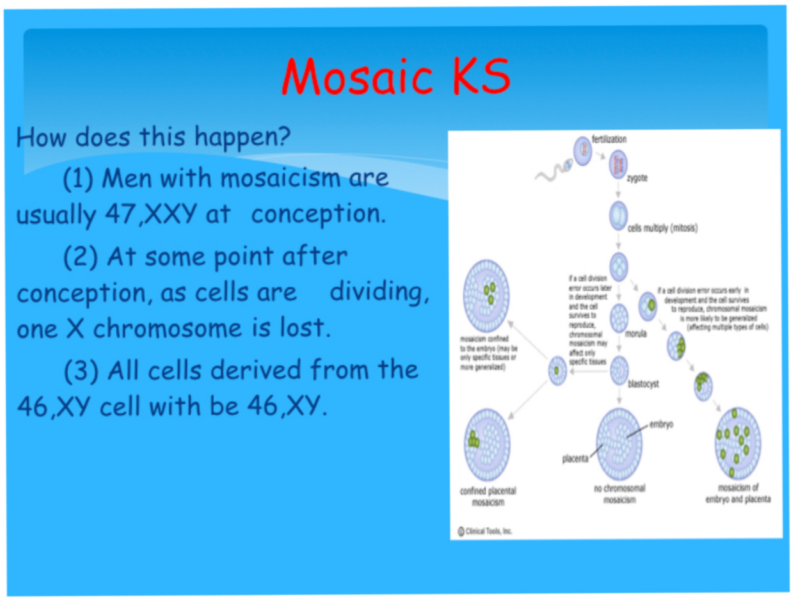
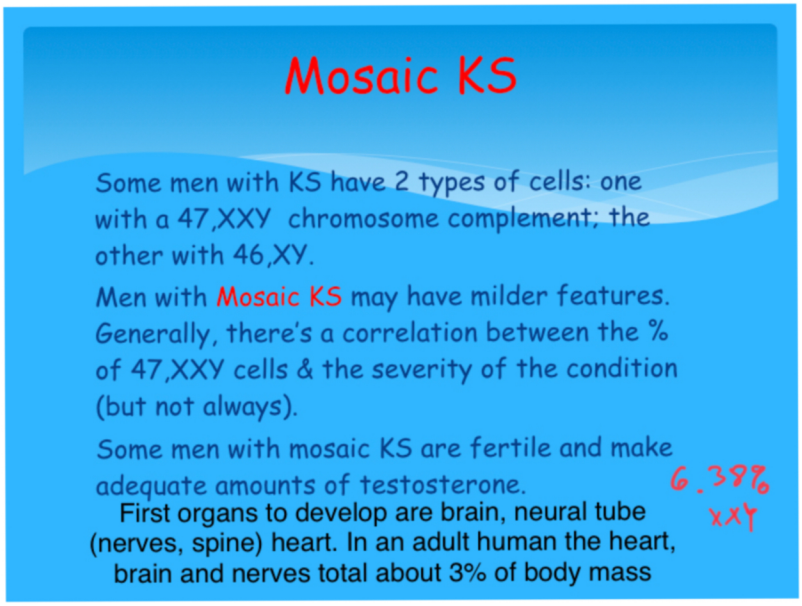


Figure 4. The figure depicts the current understanding of the genomics of KS, incorporating recent genomic results. Arrows depict possible, but not proven, pathways.

TABLE 1: Karyotypes found in the 98 patients of this study.

Karyotype	*n*	%
Pure lineage	*86*	*87.76*
47,XXY	82	83.67
48,XXXY	1	1.02
48,XXYY	3	3.06
Mosaicisms	*12*	*12.24*
47,XXY/46,XY	7	7.14
47,XXY/48,XXYY	2	2.04
47,XXY[4]/48,XXXY[2]/46,XY[44]	1	1.02
49,XXXXY[44]/48,XXXY[6]	1	1.02
47,XXY[36]/48,XXXY[1]/46,XX[1]/46,XY[2]	1	1.02





Klinefelter syndrome (KS) was first described by Harry F. Klinefelter in 1942 [Klinefelter et al., 1942]. He reported 9 men with testicular abnormalities who failed to produce sperm and had gynecomastia. In 1959, this was found to be the result of an additional X chromosome [Jacobs and Strong, 1959]. About 80% of KS patients show a 47,XXY karyotype, 20% have other numeric sex chromosome abnormalities (48,XXXY, 48,XXYY, 49,XXXXY), 46,XY/47,XXY mosaicism, or structurally abnormal sex chromosomes [Lanfranco et al., 2004].

In approximately half of the Klinefelter cases the aberrant X chromosome is thought to be paternally derived, and recent evidence suggests that it may be related to advancing paternal age, although this is controversial [Ja-

Albright's Saturday morning clinics were famous throughout the Massachusetts General Hospital. At the first one I attended, I saw a tall black boy named George Bland who had gynecomastia and very small testes (1 .O to 1.5 cm in length). When I asked Dr. Albright what this was all about, he said he did not know but that he would be happy for me to work on it. During the rest of the year, we found eight other patients with this same condition and reported the series at the endocrine meetings in 1942. Dr. Albright was charitable enough to let me put my name first on the paper that was published later in 1942 in the *Journal of Clinical Endocrinology.*' The title, "A Syndrome Characterized by Gynecomastia, Aspermatogenesis Without Aleydigism, and Increased Excretion of Follicle-Stimulating Hormone," was so long that the syndrome came to be known by my name. though it was really just another of Dr. Albright's diseases. Albright had more ideas in a day than most people have in a lifetime, and it was a great pleasure and privilege to work with him. Not only did he have great ideas and theories, but if someone came up with a fact that blasted his current theory, he soon had another one!