Development of mammalian sex organs is remarkable process. Both ovaries and testes rise from the same precursor but then differentiate into morphologically and functionally different structure. The determining factors that decide the fate of undifferentiated structures are not just the genes themselves but also the timing of transcription regulatory genes and specific amount of their products. Development of male and female gonads manages a large set of regulatory genes that interact with each other. Together it forms the gene regulatory network. Crucial role in male sex differentiation plays an SRY gene which regulates the time and quantity of the other factors expression. Even slight errors (mutations) in genes sequences of regulatory genes could change their expression. This leads to disruption of the development of sex organs or even death of the whole organism. Naturally occurring mutations and their phenotype in humans are suitable for studies of gene sex regulatory factors. Thanks to these studies we are able to detect the cause of many birth defects what is a first step toward their eventual treatment.