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Partial SHOX duplications associated with various cases of congenital uterovaginal aplasia (MRKH syndrome): A tangible evidence but a puzzling mechanism

The Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is the most severe form of congenital malformation of the inner female reproductive tract. It is diagnosed as such when the uterus, the upper vagina and optionally the Fallopian tubes are absent. It accounts for approximately 1 in 5000 live-born females and has been classified in two subtypes: type 1 in the presence of isolated uterovaginal aplasia and type 2 when associated in various combinations with extragenital malformations of the kidneys, skeleton, heart and auditory system. Most cases of MRKH syndrome are sporadic, although a significant number of many familial cases have been reported to date. Despite numerous studies, the genetics of the syndrome remains largely unknown and appears to be heterogeneous: chromosomal abnormalities and some candidate gene variants appear to be associated with a few cases; others have been suggested but not yet confirmed. To date, mainly the GREB1L gene appears to be a serious candidate. Among the remaining hypotheses, the controversial contribution of partial duplications of the SHOX gene is still puzzling, as the deficiency of this gene is a major cause of skeletal adysplasia syndromes. We have attempted to resolve this controversy in a study of 60 MRKH cases. Our results tend to show that SHOX duplications can be the origin of a genetic mechanism responsible for MRKH syndrome.