

Fraser syndrome is a rare genetic disorder characterized by partial webbing of the fingers and/or toes (partial syndactyly), kidney (renal) abnormalities, genital malformations, and/or, in some cases, complete fusion of the eyelids (cryptophthalmos) that may be associated with malformation of the eyes, causing blindness. In infants with Fraser syndrome, renal malformations may include improper development (dysplasia), underdevelopment (hypoplasia), or absence of one or both kidneys (unilateral or bilateral renal agenesis). In affected males, one or both testes may fail to descend into the scrotum (cryptorchidism), the urinary opening (meatus) may be abnormally placed on the underside of the penis (hypospadias), and/or the penis may be abnormally small (micropenis). Affected females may have malformed fallopian tubes, an abnormally enlarged clitoris (clitoromegaly), and/or an abnormally shaped uterus (bicornate uterus). In addition, the folds of skin on either side of the vaginal opening (labia) may be abnormally fused. Infants and children with Fraser syndrome may also have additional abnormalities including malformations of the middle and outer ear that may result in hearing impairment. Fraser syndrome is inherited as an autosomal recessive genetic trait. Fraser syndrome affects males and females in equal numbers. Diagnosis depends on a good family history and physical examination. Confirmation is generally easy and is performed by means of imaging devices.