

X-linked ichthyosis is a genetic skin disorder that affects males. It is an inborn error of metabolism characterized by a deficiency of the enzyme steroid sulfatase. Under normal conditions, this enzyme breaks down (metabolizes) cholesterol sulfate, a member of the chemical family of steroids. Cholesterol sulfate plays a role in maintaining the integrity of the skin. If steroid metabolism is interrupted and cholesterol sulfate accumulates in the skin cells, the skin cells stick together more strongly than usual. The normal shedding of dead skin cells is inhibited and the skin cells build up and clump into scales. X-linked recessive disorders are conditions that are coded on the X chromosome. Females have two X chromosomes; males have one X chromosome and one Y chromosome. Therefore, in females, the normal gene on one X chromosome can mask disease traits on the other X chromosome. Since males have only one X chromosome, if they inherit the gene for a disease present on the X they will express the disease. Men with X-linked disorders transmit the gene to all their daughters, who are carriers, but never to their sons. Women who are carriers of an X-linked disorder have a 50 percent chance of transmitting the carrier condition to their daughters and a 50 percent risk of transmitting the disease to their sons. X-linked ichthyosis is a rare disorder affecting one in 6,000 males.