EBP Gene

Subjects: Genetics & Heredity
Contributor: Vivi Li

EBP, cholestenol delta-isomerase

Keywords: genes

1. Normal Function

The *EBP* gene provides instructions for making an enzyme called 3β-hydroxysteroid-Δ8,Δ7-isomerase. This enzyme is responsible for one of the final steps in the production of cholesterol. Specifically, it converts a molecule called 8(9)-cholestenol to lathosterol. Other enzymes then modify lathosterol to produce cholesterol.

Cholesterol is a waxy, fat-like substance that is produced in the body and obtained from foods that come from animals (particularly egg yolks, meat, poultry, fish, and dairy products). Although too much cholesterol is a risk factor for heart disease, this molecule is necessary for normal embryonic development and has important functions both before and after birth. It is a structural component of cell membranes and plays a role in the production of certain hormones and acids used in digestion (bile acids).

2. Health Conditions Related to Genetic Changes

2.1 X-linked Chondrodysplasia Punctata 2

More than 55 mutations in the *EBP* gene have been found to cause X-linked chondrodysplasia punctata 2, a condition that occurs almost exclusively in females and is characterized by bone, skin, and eye abnormalities. Some of the mutations responsible for this condition in females insert or delete a small amount of genetic material from the *EBP* gene, while others change single protein building blocks (amino acids) in the 3β-hydroxysteroid-Δ8,Δ7-isomerase enzyme. All of these mutations impair the normal function of the enzyme, preventing cells from producing enough cholesterol. A shortage of this enzyme also allows potentially toxic byproducts of cholesterol production to build up in the body. The combination of low cholesterol levels and an accumulation of other substances likely disrupts the growth and development of many body systems. It is not known, however, how this disturbance in cholesterol production leads to the specific features of X-linked chondrodysplasia punctata 2.

Rarely, a severe form of X-linked chondrodysplasia punctata 2 has been reported in males. These cases result from changes involving single amino acids in the 3β-hydroxysteroid-Δ8,Δ7-isomerase enzyme. Affected males have some of the same features as affected females, as well as changes in the structure of the brain, moderately to profoundly delayed development, and other birth defects.

3. Other Names for This Gene

- 3-beta-hydroxysteroid-Delta(8),Delta(7)-isomerase
- 3-beta-hydroxysteroid-delta-8,delta-7-isomerase
- CDPX2
- CPXD
- D8-D7 sterol isomerase
- delta(8)-Delta(7) sterol isomerase
- EBP_HUMAN

- emopamil binding protein (sterol isomerase)
- emopamil-binding protein (sterol isomerase)
- sterol 8-isomerase

References

1. Ausavarat S, Tanpaiboon P, Tongkobpetch S, Suphapeetiporn K, Shotelersuk V.Two novel EBP mutations in Conradi-Hünermann-Happle syndrome. Eur J Dermatol.2008 Jul-Aug;18(4):391-3. doi: 10.1684/ejd.2008.0433.
2. Braverman N, Lin P, Moebius FF, Obie C, Moser A, Glossmann H, Wilcox WR,Rimoin DL, Smith M, Kratz L, Kelley RI, Valle D. Mutations in the gene encoding 3beta-hydroxysteroid-delta 8, delta 7-isomerase cause X-linked dominantConradi-Hünermann syndrome. Nat Genet. 1999 Jul;22(3):291-4.
3. Furtado LV, Bayrak-Toydemir P, Hulinsky B, Damjanovich K, Carey JC, Rope AF. Anovel X-linked multiple congenital anomaly syndrome associated with an EBPmutation. Am J Med Genet A. 2010 Nov;152A(11):2838-44. doi: 10.1002/ajmg.a.33674.
4. Herman GE, Kelley RI, Pureza V, Smith D, Kopacz K, Pitt J, Sutphen R,Sheffield LJ, Metzenberg AB. Characterization of mutations in 22 females withX-linked dominant chondrodysplasia punctata (Happle syndrome). Genet Med. 2002Nov-Dec;4(6):434-8.
5. Ikegawa S, Ohashi H, Ogata T, Honda A, Tsukahara M, Kubo T, Kimizuka M,Shimode M, Hasegawa T, Nishimura G, Nakamura Y. Novel and recurrent EBP mutationsin X-linked dominant chondrodysplasia punctata. Am J Med Genet. 2000 Oct2;94(4):300-5. Review.
6. Milunsky JM, Maher TA, Metzenberg AB. Molecular, biochemical, and phenotypicanalysis of a hemizygous male with a severe atypical phenotype for X-linkeddominant Conradi-Hunermann-Happle syndrome and a mutation in EBP. Am J Med Genet A. 2003 Jan 30;116A(3):249-54.
7. Steijlen PM, van Geel M, Vreeburg M, Marcus-Soekarman D, Spaapen LJ,Castelijns FC, Willemsen M, van Steensel MA. Novel EBP gene mutations inConradi-Hünermann-Happle syndrome. Br J Dermatol. 2007 Dec;157(6):1225-9.