

## Kawasaki disease

### Description

Kawasaki disease is a sudden and time-limited (acute) illness that affects infants and young children. Affected children develop a prolonged fever lasting several days, a skin rash, and swollen lymph nodes in the neck (cervical lymphadenopathy). They also develop redness in the whites of the eyes (conjunctivitis) and redness (erythema) of the lips, lining of the mouth (oral mucosa), tongue, palms of the hands, and soles of the feet.

Without treatment, 15 to 25 percent of individuals with Kawasaki disease develop bulging and thinning of the walls of the arteries that supply blood to the heart muscle (coronary artery aneurysms) or other damage to the coronary arteries, which can be life-threatening.

### Frequency

In the United States and other Western countries, Kawasaki disease occurs in approximately 1 in 10,000 children under 5 each year. The condition is 10 to 20 times more common in East Asia, including Japan, Korea, and Taiwan.

### Causes

The causes of Kawasaki disease are not well understood. The disorder is generally regarded as being the result of an abnormal immune system activation, but the triggers of this abnormal response are unknown. Because cases of the disorder tend to cluster geographically and by season, researchers have suggested that an infection may be involved. However, no infectious agent (such as a virus or bacteria) has been identified.

A variation in the *ITPKC* gene has been associated with an increased risk of Kawasaki disease. The *ITPKC* gene provides instructions for making an enzyme called inositol 1,4,5-trisphosphate 3-kinase C. This enzyme helps limit the activity of immune system cells called T cells. T cells identify foreign substances and defend the body against infection. Reducing the activity of T cells when appropriate prevents the overproduction of immune proteins called cytokines that lead to inflammation and which, in excess, cause tissue damage. Researchers suggest that the *ITPKC* gene variation may interfere with the body's ability to reduce T cell activity, leading to inflammation that damages blood vessels and results in the signs and symptoms of Kawasaki disease.

It appears likely that other factors, including changes in other genes, also influence the development of this complex disorder.

#### Learn more about the gene associated with Kawasaki disease

- ITPKC

### **Inheritance**

A predisposition to Kawasaki disease appears to be passed through generations in families, but the inheritance pattern is unknown. Children of parents who have had Kawasaki disease have twice the risk of developing the disorder compared to the general population. Children with affected siblings have a tenfold higher risk.

### **Other Names for This Condition**

- Acute febrile mucocutaneous lymph node syndrome
- Kawasaki syndrome
- KD
- Mucocutaneous lymph node syndrome

### **Additional Information & Resources**

#### Genetic Testing Information

- Genetic Testing Registry: Acute febrile mucocutaneous lymph node syndrome (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0026691/>)

#### Genetic and Rare Diseases Information Center

- Kawasaki disease (<https://rarediseases.info.nih.gov/diseases/6816/index>)

#### Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

#### Clinical Trials

- ClinicalTrials.gov ([https://clinicaltrials.gov/search?cond=%22Kawasaki disease%22](https://clinicaltrials.gov/search?cond=%22Kawasaki+disease%22))

#### Catalog of Genes and Diseases from OMIM

- KAWASAKI DISEASE (<https://omim.org/entry/611775>)

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28Mucocutaneous+Lymph+Node+Syndrome%5BMAJR%5D%29+AND+%28Kawasaki+disease%5BTI%5D%29+AND+review%5Bpt%5D+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>)

### **References**

- Burns JC. Kawasaki Disease update. *Indian J Pediatr.* 2009 Jan;76(1):71-6. doi:10.1007/s12098-009-0031-3. Epub 2009 Apr 18. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19391005>)
- Harnden A, Takahashi M, Burgner D. Kawasaki disease. *BMJ.* 2009 May5;338:b1514. doi: 10.1136/bmj.b1514. No abstract available. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19416993>)
- Hata A, Onouchi Y. Susceptibility genes for Kawasaki disease: toward implementation of personalized medicine. *J Hum Genet.* 2009 Feb;54(2):67-73. doi:10.1038/jhg.2008.9. Epub 2009 Jan 16. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19158812>)
- Kuo HC, Yang KD, Juo SH, Liang CD, Chen WC, Wang YS, Lee CH, Hsi E, Yu HR, Woon PY, Lin IC, Huang CF, Hwang DY, Lee CP, Lin LY, Chang WP, Chang WC. ITPKC single nucleotide polymorphism associated with the Kawasaki disease in a Taiwanese population. *PLoS One.* 2011 Apr 14;6(4):e17370. doi:10.1371/journal.pone.0017370. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/21533171>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3077380/>)
- Lin MT, Wang JK, Yeh JI, Sun LC, Chen PL, Wu JF, Chang CC, Lee WL, Shen CT, Wang NK, Wu CS, Yeh SZ, Chen CA, Chiu SN, Wu MH. Clinical Implication of the C Allele of the ITPKC Gene SNP rs28493229 in Kawasaki Disease: Association With Disease Susceptibility and BCG Scar Reactivation. *Pediatr Infect Dis J.* 2011 Feb;30(2):148-52. doi: 10.1097/INF.0b013e3181f43a4e. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/20805785>)
- Onouchi Y, Gunji T, Burns JC, Shimizu C, Newburger JW, Yashiro M, Nakamura Y, Yanagawa H, Wakui K, Fukushima Y, Kishi F, Hamamoto K, Terai M, Sato Y, Ouchi K, Saji T, Nariai A, Kaburagi Y, Yoshikawa T, Suzuki K, Tanaka T, Nagai T, Cho H, Fujino A, Sekine A, Nakamichi R, Tsunoda T, Kawasaki T, Nakamura Y, Hata A. ITPKC functional polymorphism associated with Kawasaki disease susceptibility and formation of coronary artery aneurysms. *Nat Genet.* 2008 Jan;40(1):35-42. doi: 10.1038/ng.2007.59. Epub 2007 Dec 16. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/18084290>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2876982/>)

- Onouchi Y. Molecular genetics of Kawasaki disease. *Pediatr Res*. 2009 May;65(5Pt 2):46R-54R. doi: 10.1203/PDR.0b013e31819dba60. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19190534>)
- Rowley AH, Shulman ST. Pathogenesis and management of Kawasaki disease. *Expert Rev Anti Infect Ther*. 2010 Feb;8(2):197-203. doi: 10.1586/eri.09.109. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/20109049>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2845298/>)
- Yeung RS. Kawasaki disease: update on pathogenesis. *Curr Opin Rheumatol*. 2010 Sep;22(5):551-60. doi: 10.1097/BOR.0b013e32833cf051. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/20616737>)

**Last updated September 1, 2015**