Recombinant 8 syndrome

Description

Recombinant 8 syndrome is a condition that involves complex congenital heart abnormalities, urinary tract abnormalities, moderate to severe intellectual disability, abnormal muscle tone, and a distinctive facial appearance.

The most common heart abnormalities are known as tetrology of Fallot and conotruncal defects. The characteristic facial features include a wide, square face; a thin upper lip; a downturned mouth; a small chin (micrognathia); wide-set eyes (hypertelorism); and low-set or unusually shaped ears. People with recombinant 8 syndrome may have overgrowth of the gums (gingival hyperplasia), abnormal tooth development, or an opening in the upper lip (cleft lip) with or without an opening in the roof of the mouth (cleft palate). Males with this condition frequently have undescended testes (cryptorchidism). Some affected individuals have recurrent ear infections (otitis media), hearing loss, or hand and finger differences. In individuals with recombinant 8 syndrome, the heart abnormalities can be life-threatening.

Frequency

Recombinant 8 syndrome is a rare condition; its exact incidence is unknown. Most people with this condition are of Hispanic ancestry, particularly from the San Luis Valley area of southern Colorado and northern New Mexico. Recombinant 8 syndrome is also called San Luis Valley syndrome. Only a few cases outside this population have been found.

Causes

Recombinant 8 syndrome is caused by a rearrangement of chromosome 8 that results in a deletion of a piece of the short (p) arm and a duplication of a piece of the long (q) arm. The deletion and duplication result in the recombinant 8 chromosome. On the recombinant 8 chromosome, there is one copy of each of the genes instead of the usual two on the section of chromosome 8p that is deleted; and there are three copies each of the genes on the section of chromosome 8q that is duplicated. The signs and symptoms of recombinant 8 syndrome are related to the loss and addition of genetic material on

these regions of chromosome 8. While the regions affected in recombinant chromosome 8 includes hundreds of genes, researchers are working to determine which genes play a role in the signs and symptoms of this condition.

Learn more about the chromosome associated with Recombinant 8 syndrome

- chromosome 8

Inheritance

Recombinant 8 syndrome is inherited in an autosomal dominant pattern, which means one copy of the recombinant chromosome 8 in each cell is sufficient to cause the disorder.

Most people with recombinant 8 syndrome have one parent with a change in chromosome 8 called an inversion. An inversion involves the breakage of a chromosome in two places; the resulting piece of DNA is reversed and reinserted into the chromosome. Genetic material is typically not lost as a result of this inversion in chromosome 8, so people usually do not have any related health problems. However, genetic material can be lost or duplicated when inversions are passed to the next generation. People with the chromosome 8 inversion are at of risk having a child with recombinant 8 syndrome.

Other Names for This Condition

- Rec(8) syndrome
- Recombinant chromosome 8 syndrome
- San Luis Valley syndrome

Additional Information & Resources

Genetic and Rare Diseases Information Center

- Recombinant 8 syndrome (https://rarediseases.info.nih.gov/diseases/9698/index)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- RECOMBINANT CHROMOSOME 8 SYNDROME (https://omim.org/entry/179613)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=((recombinant+8+syndrome%5BTIAB%5D)+OR+(San+Luis+Valley+syndrome%5BTIAB%5D)+%20OR+(recombinant+chromosome+8+syndrome%5BTIAB%5D)+OR+(rec+8+syndrome%5BTIAB%5D))+AND+english%5Bla%5D+AND+human%5Bmh%5D)

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