Recombinant 8 Syndrome

Subjects: Genetics & Heredity
Contributor: Nora Tang

Recombinant 8 syndrome is a condition that involves heart and urinary tract abnormalities, moderate to severe intellectual disability, and a distinctive facial appearance. 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) and abnormal tooth development. Males with this condition frequently have undescended testes (cryptorchidism). Some affected individuals have recurrent ear infections (otitis media) or hearing loss. Many children with recombinant 8 syndrome do not survive past early childhood, usually due to complications related to their heart abnormalities.

genetic conditions

1. Introduction

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) and abnormal tooth development. Males with this condition frequently have undescended testes (cryptorchidism). Some affected individuals have recurrent ear infections (otitis media) or hearing loss. Many children with recombinant 8 syndrome do not survive past early childhood, usually due to complications related to their heart abnormalities.

2. Frequency

Recombinant 8 syndrome is a rare condition; its exact incidence is unknown. Most people with this condition are descended from a Hispanic population originating in 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.

3. 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. The signs and symptoms of recombinant 8 syndrome are related to the loss and

addition of genetic material on these regions of chromosome 8. Researchers are working to determine which genes are involved in the deletion and duplication on chromosome 8.

3.1. The Chromosome Associated with Recombinant 8 Syndrome

- chromosome 8

4. Inheritance

This condition 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 at least 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 being passed to the next generation. People with this chromosome 8 inversion are at of risk having a child with recombinant 8 syndrome.

5. Other Names for This Condition

- rec(8) syndrome
- recombinant chromosome 8 syndrome
- San Luis Valley syndrome

References

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