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Recombinant 8 syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

Recombinant 8 syndrome. ORPHA:96167

Recombinant 8 (rec(8)) syndrome, also known as San Luis Valley syndrome, is a complex chromosomal disorder that is due to a parental pericentric inversion of chromosome 8 and is characterized by major congenital heart anomalies, urogenital malformations, moderate to severe intellectual deficiency and mild craniofacial dysmorphism.