# Chromoanasynthesis as a cause of Jacobsen syndrome


## Abstract

Jacobsen syndrome (MIM #147791) is a rare multisystem genomic disorder involving craniofacial abnormalities, intellectual disability, other neurodevelopmental defects, and terminal truncation of chromosome 11q, typically deleting ~170 to >340 genes. We describe the first case of Jacobsen syndrome caused by congenital chromoanasynthesis, an extreme form of complex chromosomal rearrangement. Six duplications and five deletions occurred on one copy of chromosome 11q with microhomology signatures in the breakpoint junctions, indicating an all-at-once replication-based rearrangement mechanism in a gametocyte or early post-zygotic cell. Eighteen genes were deleted from the Jacobsen region, including KIRREL3, which is associated with intellectual disability.



Anzick S, Thurm A, Burkett S, Velez D, Cho E, Chlebowski C, Virtaneva K, Bruno D, Martin CB, Lang DM, Brooks B, Martens C, McDermott DH, Murphy PM. Chromoanasynthesis as a cause of Jacobsen syndrome. Am J Med Genet A. 2020 Nov;182(11):2533-2539. doi: 10.1002/ajmg.a.61824. Epub 2020 Aug 25. PMID: 32841469.