WHITEPAPER

Biocare Basics: Ring Chromosomes

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BIOCARE MEDICAL

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Ring chromosomes are a rare type of chromosomal abnormality characterized by a circular or ring-like structure formed from the fusion of the chromosome's two ends.[1,8] This genetic anomaly can occur in any of the 22 autosomal chromosomes or the sex chromosomes (X or Y), leading to various clinical consequences depending on the specific chromosome involved and the extent of genetic material loss.[1,8] Accurate detection methods are necessary for identification and appropriate medical management.

Though the process is still not fully understood, ring chromosomes typically arise from terminal breaks on the ends of both chromosome arms, followed by the fusion of the broken ends into a circular configuration.[1,3,7,8] In other cases, two telomeric regions may spontaneously fuse with no genetic deletion involved.[1,2,7,8] This fusion can occur spontaneously during cell division or may result from damage due to radiation exposure, chemicals, or genetic mutations.

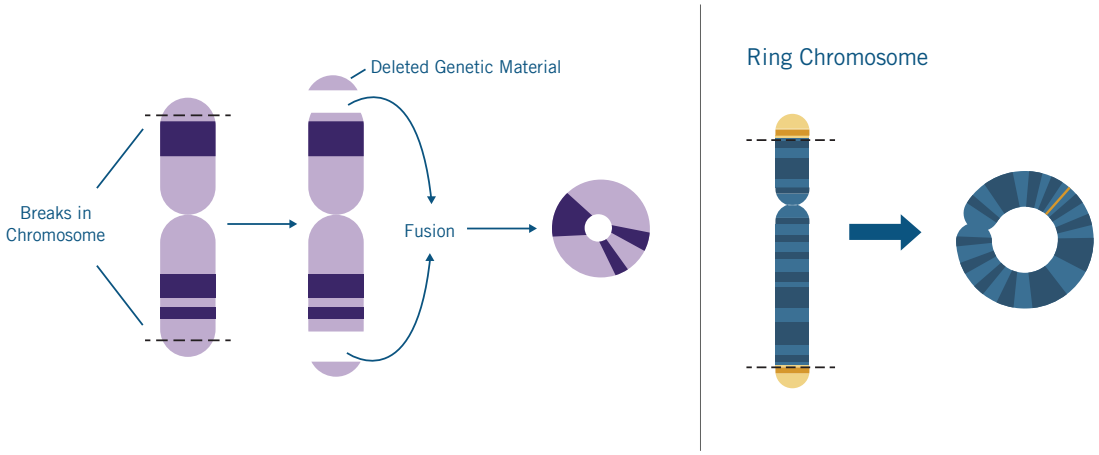
The exact mechanisms leading to ring chromosome formation can vary, and the timing of the fusion event determines how cells are affected.[1,7] Ring formation in the germ cell line will cause the ring chromosome to be present in every cell, while formation in the somatic cell line may only affect a subset of cells.[1,7] An extreme minority of cases have shown parental transmission of ring chromosomes, but this has been observed only sporadically with no known gender or ethnicity bias.[1,2,8]

The clinical significance of ring chromosomes depends on the specific chromosome involved, the size of the ring, and the extent of genetic material lost during the formation process.[1]

The effects of ring chromosomes can range from mild to severe, and the same ring chromosome may exhibit different symptoms in different individuals. Some carriers appear to show no symptoms.[2,7] However, many individuals with ring chromosomes experience developmental and intellectual disabilities.[4,5] Common features may include growth abnormalities, cognitive impairment, delayed speech and motor skills, and facial dysmorphisms.[4,5] Ring chromosome formation on chromosome 20 has been associated with epilepsy.[2,5,8]

Several testing methods are employed to detect the presence of ring chromosomes, including genome sequencing, Multiplex Ligation-Dependent Probe Amplification (MLPA), and Fluorescence In Situ Hybridization (FISH).[1,3,8] FISH uses fluorescent DNA probes that bind to specific regions of chromosomes, which may allow researchers to identify breakpoints and genomic imbalances and define the mechanism of ring formation.[3] Studies have employed RP11 BAC FISH probes to study ring chromosomes in patient blood samples.[3]

Ring Chromosome Illustration



To learn about Empire Genomics RP11 BAC FISH probes, please visit our website at empiregenomics.com, call us at 1-800-715-5880, or send us an email at info@empiregenomics.com

1. Daber, R. D., Conlin, L. K., Leonard, L. D., Canevini, M. P., Vignoli, A., Hosain, S., Brown, L. W., & Spinner, N. B. (2012). Ring chromosome 20. European Journal of Medical Genetics, 55(5), 381-387. ISSN 1769-7212. https://doi.org/10.1016/j.ejmg.2012.02.004. 2. Gardner, R. J. McKinlay, Grant R. Sutherland, and Lisa G. Shaffer, 'Autosomal ring chromosomes', Chromosome Abnormalities and Genetic Counseling, 4 edn, Oxford Monographs on Medical Genetics (2011; online edn, Oxford Academic, 1 Oct. 2012), https://doi.org/10.1093/med/9780195375336.003.0011, accessed 27 Sept. 2023. 3. Guilherme, R.S., Ayres Meloni, V.F., Kim, C.A. et al. Mechanisms of ring chromosome formation, ring instability and clinical consequences. BMC Med Genet 12, 171 (2011). https://doi.org/10.1186/1471-2350-12-171 4. Jeffries, A.R., Curran, S., Elmslie, F., Sharma, A., Wenger, S., Hummel, M. and Powell, J. (2005), Molecular and phenotypic characterization of ring chromosome 22. Am. J. Med. Genet., 137A: 139-147. https://doi.org/10.1002/ajmg.a.30780 5. MedlinePlus. (2009, May 1). Ring Chromosome 14 Syndrome. In MedlinePlus [Internet]. Bethesda, MD: National Library of Medicine (US). https://medlineplus.gov/genetics/condition/ring-chromosome-14-syndrome/ (Updated Oct 1, 2015; Oct 1, 2015; Accessed Sept 28, 2023). 6. MedlinePlus. (2009, May 1). Ring Chromosome 20 Syndrome. In MedlinePlus [Internet]. Bethesda, MD: National Library of Medicine (US). https://medlineplus.gov/genetics/condition/ring-chromosome-20-syndrome/ (Updated May 1, 2009; Reviewed May 1, 2009; Accessed Sept 28, 2023). 7. Pristyazhnyuk, I.E., Menzorov, A.G. Ring chromosomes: from formation to clinical potential. Protoplasma 255, 439–449 (2018). https://doi.org/10.1007/s00709-017-1165-1 8. Yip M. Y. (2015). Autosomal ring chromosomes in human genetic disorders. Translational pediatrics, 4(2), 164–174. https://doi.org/10.3978/j.issn.2224-4336.2015.03.04