

Translocations that appear balanced karyotypically often show imbalance by sequencing. For example, a balanced translocation between chromosomes 11 and 12 (11;12) may appear to be balanced karyotypically, but sequencing reveals a loss of heterozygosity at the *TP53* locus on chromosome 11. This loss of heterozygosity is due to a deletion of the *TP53* gene on chromosome 11, which is located on the derivative chromosome 11. This loss of heterozygosity is described as a loss of heterozygosity (LOH) at the *TP53* locus.

17 References

- Al-Aish MS: Human chromosome morphology. I. Studies on normal chromosome characterization, classification and karyotyping. *Can J Genet Cytol* 11:370–381 (1969).
- Baliakas P, Jeromin S, Iskas M, Puiggros A, Plevova K, Nguyen-Khac F, Davis Z, Rigolin GM, Visentin A, Xochelli A, et al.: Cytogenetic complexity in chronic lymphocytic leukemia: definitions, associations, and clinical impact. *Blood* 133:1205–1216 (2019).
- Caspersson T, Farber S, Foley GE, Kudynoski J, Modest EJ, Simonsson E, Wagh U, Zech L: Chemical differentiation along metaphase chromosomes. *Exp Cell Res* 49:219–222 (1968).
- Caspersson T, Lomakka G, Zech L: The 24 fluorescence patterns of human metaphase chromosomes – distinguishing characters and variability. *Hereditas* 67:89–102 (1972).
- Chicago Conference (1966): Standardization in Human Cytogenetics. *Birth Defects: Original Article Series*, Vol 2, No 2 (The National Foundation, New York 1966).
- Chun K, Hagemeijer A, Iqbal A, Slovak ML: Implementation of standardized international karyotype scoring practices is needed to provide uniform and systematic evaluation for patients with myelodysplastic syndrome using IPSS criteria: An International Working Group on MDS Cytogenetics Study. *Leukemia Res* 34:160–165 (2010).
- Cremer T, Landegent J, Bruckner A, Scholl HP, Schardin M, Hager HD, Devilee P, Pearson P, van der Ploeg M: Detection of chromosome aberrations in the human interphase nucleus by visualization of specific target DNAs with radioactive and non-radioactive *in situ* hybridization techniques: diagnosis of trisomy 18 with probe L1.84. *Hum Genet* 74:346–352 (1986).
- den Dunnen JT, Dalgleish R, Maglott DR, Hart RK, Greenblatt MS, McGowan-Jordan J, Roux AF, Smith T, Antonarakis SE, Taschner PE: HGVS recommendations for the description of sequence variants: 2016 update. *Hum Mutat* 37:564–569 (2016).
- Denver Conference (1960): A proposed standard system of nomenclature of human mitotic chromosomes. *Lancet* i:1063–1065 (1960).
- Dutrillaux B: Obtention simultanée de plusieurs marquages chromosomiques sur les mêmes préparations, après traitement par le BrdU. *Humangenetik* 30:297–306 (1975).
- Ford CE, Hamerton JL: The chromosomes of man. *Nature* 178:1020–1023 (1956).
- Francke U: High-resolution ideograms of trypsin-Giemsa banded human chromosomes. *Cytogenet Cell Genet* 31:24–32 (1981).
- Francke U: Digitized and differentially shaded human chromosome ideograms for genomic applications. *Cytogenet Cell Genet* 65:206–218 (1994).
- Francke U, Oliver N: Quantitative analysis of high-resolution trypsin-Giemsa bands on human prometaphase chromosomes. *Hum Genet* 45:137–165 (1978).
- Grimwade D, Hills RK, Moorman AV, Walker H, Chatters S, Goldstone AH, Wheatley K, Harrison CJ, Burnett AK, National Cancer Research Institute Adult Leukaemia Working Group: Refinement of cytogenetic classification in acute myeloid leukemia: determination of prognostic significance of rare recurring chromosomal abnormalities among 5876 younger adult patients treated in the United Kingdom Medical Research Council trials. *Blood* 116:345–365 (2010).
- Guan XY, Meltzer PS, Trent JM: Rapid generation of whole chromosome painting probes (WCPs) by chromosome microdissection. *Genomics* 22:101–107 (1994).
- Haase D, Stevenson KE, Neuberg D, Maciejewski JP, Nazha A, Sekeres MA, Ebert BL, Garcia-Manero G, Haferlach C, Haferlach T, et al.: *TP53* mutation status divides myelodysplastic syndromes with complex karyotypes into distinct prognostic subgroups. *Leukemia* 33:1747–1758 (2019).

- ISCN (1978): An International System for Human Cytogenetic Nomenclature. Birth Defects: Original Article Series, Vol 14, No 8 (The National Foundation, New York 1978); also in *Cytogenet Cell Genet* 21:309–404 (1978).
- ISCN (1981): An International System for Human Cytogenetic Nomenclature – High Resolution Banding. Birth Defects: Original Article Series, Vol 17, No 5 (March of Dimes Birth Defects Foundation, New York 1981); also in *Cytogenet Cell Genet* 31:1–23 (1981).
- ISCN (1985): An International System for Human Cytogenetic Nomenclature, Harnden DG, Klinger HP (eds), Birth Defects: Original Article Series, Vol 21, No 1 (March of Dimes Birth Defects Foundation, New York 1985).
- ISCN (1991): Guidelines for Cancer Cytogenetics, Supplement to An International System for Human Cytogenetic Nomenclature, Mitelman F (ed), (S Karger, Basel 1991).
- ISCN (1995): An International System for Human Cytogenetic Nomenclature, Mitelman F (ed), (S Karger, Basel 1995).
- ISCN (2005): An International System for Human Cytogenetic Nomenclature, Shaffer LG, Tommerup N (eds), (S Karger, Basel 2005).
- ISCN (2009): An International System for Human Cytogenetic Nomenclature, Shaffer LG, Slovak ML, Campbell LJ (eds), (S Karger, Basel 2009).
- ISCN (2013): An International System for Human Cytogenetic Nomenclature, Shaffer LG, McGowan-Jordan J, Schmid M (eds), (S Karger, Basel 2012).
- ISCN 2016: An International System for Human Cytogenomic Nomenclature (2016); McGowan-Jordan J, Simons A, Schmid M (eds), (Karger, Basel 2016); also in *Cytogenet Genome Res* 149:1–140 (2016).
- Jhanwar SC, Burns JP, Alonso ML, Hew W, Chaganti RSK: Mid-pachytene chromomere maps of human autosomes. *Cytogenet Cell Genet* 33:240–248 (1982).
- Kallioniemi A, Kallioniemi OP, Sudar D, Rutovitz D, Gray JW, Waldman F, Pinkel D: Comparative genomic hybridization for molecular cytogenetic analysis of solid tumors. *Science* 258:818–821 (1992).
- Landegent JE, Jansen in de Wal N, Dirks RW, Baao F, van der Ploeg M: Use of whole cosmid cloned genomic sequences for chromosomal localization by non-radioactive *in situ* hybridization. *Hum Genet* 77:366–370 (1987).
- Levan A, Frega K, Sandberg AA: Nomenclature for centromeric position on chromosomes. *Hereditas* 52:201–220 (1964).
- Lichter P, Cremer T, Borden J, Manuelidis L, Ward DC: Delineation of individual human chromosomes in metaphase and interphase cells by *in situ* suppression hybridization using recombinant DNA libraries. *Hum Genet* 80:224–234 (1988).
- Lichter P, Tang CJ, Call K, Hermanson G, Evans GA, Housman D, Ward DC: High-resolution mapping of human chromosome 11 by *in situ* hybridization with cosmid clones. *Science* 247:64–69 (1990).
- Lehr T, Claussen U, Starke H: Small supernumerary marker chromosomes (sSMC) in humans. *Cytogenet Genome Res* 107:55–67 (2004).
- Lehr T, Starke H, Heller A, Kosyakova N, Mrasek K, Gross M, Karst C, Steinhaeuser U, Hunstig F, Fickelscher I, et al: Multicolor fluorescence *in situ* hybridization (FISH) applied to FISH-banding. *Cytogenet Genome Res* 114:240–244 (2006).
- London Conference on the Normal Human Karyotype. *Cytogenetics* 2:264–268 (1963).
- Magenis RE, Barton SJ: Delineation of human prometaphase paracentromeric regions using sequential GTG- and C-banding. *Cytogenet Cell Genet* 45:132–140 (1987).
- Newman S, Hermetz KE, Weckselblatt B, Rudd K: Next-generation sequencing of duplication CNVs reveals that most are tandem and some create fusion genes at breakpoints. *Am J Hum Genet* 96:208–220 (2015).
- Ordulu Z, Wong KE, Currall BB, Ivanov AR, Pereira SA, Gusella JF, Talkowski ME, Morton CC: Describing sequencing results of structural chromosome rearrangements with a suggested next-generation cytogenetic nomenclature. *Am J Hum Genet* 94:1–15 (2014).
- Paris Conference (1971): Standardization in Human Cytogenetics. Birth Defects: Original Article Series, Vol 8, No 7 (The National Foundation, New York 1972); also in *Cytogenetics* 11:313–362 (1972).
- Paris Conference (1971), Supplement (1975): Standardization in Human Cytogenetics. Birth Defects: Original Article Series, Vol 11, No 9 (The National Foundation, New York 1975); also in *Cytogenet Cell Genet* 15: 201–238 (1975).
- Parra I, Windle B: High resolution visual mapping of stretched DNA by fluorescent hybridization. *Nat Genet* 5:17–21 (1993).
- Patau K: The identification of individual chromosomes, especially in man. *Am J Hum Genet* 12:250–276 (1960).
- Pinkel D, Landegent J, Collins C, Fuscoe J, Segraves R, Lucas J, Gray JW: Fluorescence *in situ* hybridization with human chromosome-specific libraries: detection of trisomy 21 and translocations of chromosome 4. *Proc Natl Acad Sci USA* 85:9138–9142 (1988).

- Schluth-Bolard C, Labalme A, Cordier M-P, Till M, Nadeau G, Tevissen H, Lesda G, Boutry-Kryza N, Rossignol S, Rocas D, et al: Breakpoint mapping by next generation sequencing reveals causative gene disruption in patients carrying apparently balanced chromosome rearrangements with intellectual deficiency and/or congenital malformations. *J Med Genet* 50:144–150 (2013).

Stephens PJ, Greenman CD, Fu B, Yang F, Bignell GR, Mudie LJ, Pleasance ED, Lau KW, Beare D, Stebbings LA, et al: Massive genomic rearrangement acquired in a single catastrophic event during cancer development. *Cell* 144:27–40 (2011).

Tjio JH, Levan A: The chromosome number of man. *Hereditas* 42:1–16 (1956).

Trask BJ: Fluorescence *in situ* hybridization: applications in cytogenetics and gene mapping. *Trends Genet* 7: 149–154 (1991).

Viegas-Pequignot E, Dutrillaux B: Une méthode simple pour obtenir des prophases et des prometaphases. *Ann Genet* 21:122–125 (1978).

Wiegant J, Kalle W, Mullenders L, Brookes S, Hoovers JM, Dauwerse JG, van Ommen GJ, Raap AK: High-resolution *in situ* hybridization using DNA halo preparations. *Hum Mol Genet* 1:587–591 (1992).

Wiegant J, Wiesmeijer CC, Hoovers JM, Schuurings E, d’Azzo A, Vrolijk J, Tanke HJ, Raap AK: Multiple and sensitive fluorescence *in situ* hybridization with rhodamine-, fluorescein-, and coumarin-labeled DNAs. *Cytogenet Cell Genet* 63:73–76 (1993).

Wyandt HE, Tonk VS (eds): *Atlas of Human Chromosome Heteromorphisms* (Springer, New York 2008).

Yunis JJ: High resolution of human chromosomes. *Science* 191:1268–1270 (1976).

Yunis JJ, Sawyer JR, Ball DW: The characterization of high-resolution G-banded chromosomes of man. *Chromosoma* 67:293–307 (1978).