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Erratum

In "ISCN 2020: An International System for Human Cytogenomic Nomenclature" [Cytogenet Genome Res. 2020;160(7–8). Doi 10.1159/000510090 and 10.1159/isbn.978-3-318-06867-2] by McGowan-Jordan J, Hastings RJ, Moore S (editors), the following corrections to the text should be observed.

In Chapter 3: Symbols, Abbreviated Terms, and General Principles, "Uniparental disomy (8.4, 14.2.1)" should correctly refer to "Uniparental disomy (See hmz 14.2.6)"

In Chapter 9.2.3: Derivative Chromosomes, the example:
"47,XX,t(9;22;6)(q34;q11.2;p21),+der(22)t(9;22;6)" should correctly read:
"47,XX,t(6;9;22)(p21;q34;q11.2),+der(22)t(6;9;22)."
In Chapter 9.2.3: Derivative Chromosomes, the example
"46,XY,der(9)t(9;22)(q34;q11),+22,der(22;22)(22pter→22q11::9q34→9qter::9qter→9q34::22q11→22pter)
An isochromosome for the derivative chromosome 22 generated by a t(9;22), i.e., an isochromosome for the long arm of a Ph chromosome" should correctly read:
"47,XY,der(9)t(9;22)(q34;q11.2),+22,der(22;22)(22pter→22q11.2::9q34→9qter::9qter→9q34::22q11.2→22pter).
An isodicentric chromosome generated from a der(22)t(9;22)."

In Chapter 9.2.5: Duplications, the example:
"46,XX,dup(1)(pter→p31::p31→p34::p31→qter) or dup(1)(pter→p34::p31→p34::p31→pter)" should correctly read:
"46,XX,dup(1)(pter→p31::p31→p34::p31→qter) or dup(1)(pter→p34::p31→p34::p34→qter)."

In Chapter 9.2.8: Homogeneously Staining Regions, the example:
"46,XX,der(1)ins(1;7)(q21;p21p11.2)hsr(1;7)(q21;p11.2)
46,XX,der(1)(1pter→1q21::hsr::7p11.2→7p21::1q21→1qter)" should correctly read:
"46,XX,der(1)ins(1;7)(q21;p21p11.2)hsr(1;7)(q21;p11.2)
46,XX,der(1)(1pter→1q21::hsr::7p21→7p11.2::1q21→1qter."

In Chapter 9.2.8: Homogeneously Staining Regions, the example:
"46,XX,der(1)ins(1;7)(q21;p21p11.2)hsr(1;7)(q21;p21)
46,XX,der(1)(1pter→1q21::7p11.2→7p21::hsr::1q21→1qter)" should correctly read:
"46,XX,der(1)ins(1;7)(q21;p21p11.2)hsr(1;7)(q21;p21)
46,XX,der(1)(1pter→1q21::7p21→7p11.2::hsr::1q21→1qter)."

In Chapter 9.2.9: Insertions, the sentence:
"The orientation of the inserted segment is indicated by the order of the bands of the inserted segment with respect to the centromere" should correctly read:

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"The orientation of the inserted segment is indicated by the order of the bands of the inserted segment from pter to qter."

In Chapter 9.2.17.1: Reciprocal Translocations, the example:
"46,XX,t(5;14;9)(q13q23;q24q21;p12p23)
46,XX,t(5;14;9)(5pter→5q13::9p12→9p23::5q23→5qter;14pter→14q21::5q13→5q23::14q24→14qter;9pter→9p23::14q21→14q24::9p12→9qter)" should correctly read:
"46,XX,t(5;14;9)(q13q23;q24q21;p12p23)
46,XX,t(5;14;9)(5pter→5q13::9p12→9p23::5q23→5qter;14pter→14q21::5q13→5q23::14q24→14qter;9pter→9p23::14q24→14q21::9p12→9qter)."

In Chapter 11.1.4: Stemline, Sideline and Clonal Evolution, the example:
"50,sl,+6,–inv(6),+7,+8,+9,+mar[11] or 50,idem,+6,–inv(6),+7,+8,+9,+mar[11]" should correctly read:
"51,sl,+6,–inv(6),+7,+8,+9,+mar[11] or 51,idem,+6,–inv(6),+7,+8,+9,+mar[11]."

In Chapter 13.2.3: Abnormal Signal Patterns with Multiple Probes, the example:
"46,X,?i(Y)(p10).ish idic(Y)(q11)(DYZ3+,DYZ1-,DYZ3+)
A presumed isochromosome for the short arm of Y was shown by ish to have two centromeres and no heterochromatin" should correctly read:
"46,X,?i(Y)(p10).ish idic(Y)(q11.1)(DYZ3+,DYZ1-,DYZ3+)
A presumed isochromosome for the short arm of Y was shown by ish to have two centromeres and no heterochromatin."

In Chapter 13.3.3: Abnormal Interphase Signal Pattern, the example:
"nuc ish(DXZ1,DYZ3)×1[34/50]/(DXZ1×1,DYZ3×0)[12/50]/(DXZ1×1,DYZ3×2)[6/50]
One copy of X and one copy of Y in 34 of 50 nuclei, in addition to 12 nuclei with a single X and six nuclei with one X and two Y chromosomes" should correctly read:
"nuc ish(DXZ1,DYZ3)×1[32/50]/(DXZ1×1,DYZ3×0)[12/50]/(DXZ1×1,DYZ3×2)[6/50]
One copy of X and one copy of Y in 32 of 50 nuclei, in addition to 12 nuclei with a single X and six nuclei with one X and two Y chromosomes."

In Chapter 14.1: Introduction, the sentence:
"Microarray nomenclature includes the genomic coordinates for banded chromosomes which are defined in the translation tables provided by NCBI (hg19/GRCh38, http://hgdownload.cse.ucsc.edu/goldenPath/hg19/database/cytoBand.txt.gz and hg38/GRCh38, http://hgdownload.cse.ucsc.edu/goldenPath/hg38/database/cytoBand.txt.gz)" should correctly read:
"Microarray nomenclature includes the genomic coordinates for banded chromosomes which are defined in the translation tables provided by NCBI (hg19/GRCh37, http://hgdownload.cse.ucsc.edu/goldenPath/hg19/database/cytoBand.txt.gz and hg38/GRCh38, http://hgdownload.cse.ucsc.edu/goldenPath/hg38/database/cytoBand.txt.gz)."

In Chapter 14.2.4: Multiple Techniques, the example:
"46,X,der(Y)t(X;Y)(p22.33;q12).arr[GRCh37] Xp22.33(701_2,679,502)×3,Xp22.33p22.2(2,709,521_15,955,588)×2,Yq11.221q11.23(16,139,805_27,177,529)×0" should be written as:
"The array defines the Yq breakpoint as q11.221. As the karyotype is given, and the abnormal chromosome is known to be a der(Y), it should be written as: 46,X,der(Y)t(X;Y)(p22.33;q11.221).arr[GRCh37] Xp22.33p22.2 (701_15,955,588)×2,Yq11.221q11.23(16,139,805_27,177,529)×0"

In Chapter 14.2.8: Polar Bodies, the following example:
"arr cht(13,14)×2,(18)×0,(21)×2" should correctly read:
"arr cht(13,14)×2,cht(18)×0,cht(21)×2."

In Chapter 15.2: Examples of RSA Nomenclature for Normal and Aneuploidy, the example:
"rsa(X)×2,(13,18)×2,(21)×2~3" should correctly read:
"rsa(X,13,18)x2,(21)x2~3."

These corrections are displayed in line in ISCN Online. Please visit https://iscn.karger.com/ to access and view the Erratum in context as well as Addenda relating to new developments.