Monitoring Group (MG) Condition	BPA Code	BPA Condition	Examples of written karyotypes and comments
Down Syndrome/Trisomy 21	Q90	Down's syndrome	
	Q90.0	Trisomy 21, meiotic nondisjunction	47,XX,+21 47,XY,+21
	Q90.1	Trisomy 21, mosaicism (mitotic nondisjunction)	46,XX / 47,XX,+21 47,XY,+21 MOS 46,XY[5]/47,XY,+21[5]
	Q90.2	Trisomy 21, translocation	Unbalanced Robertsonian Translocation - trisomy 21 46,XY,der(14;21)(q10;q10),+21 **NOT** 45,XX,der(21;21)(q10;q10) which is balanced
	Q90.9	Down's syndrome, unspecified Trisomy 21 NOS	nuc ish 21q22.13(D21S259x3) 46,XY,dup(21)(q22.1q22.3)
	Q91.0	Trisomy 18, meiotic nondisjunction	Edward's Syndrome 47,XX,+18 47,XY,+18
Edwards' syndrome/Trisomy 18	Q91.0-Q91.3		
	Q91.1	Trisomy 18, mosaicism (mitotic nondisjunction)	47,XX,+18/46,XX 46,XY[10]/47,XY,+18[40]
	Q91.2	Trisomy 18, translocation	
	Q91.3	Edwards' syndrome, unspecified	nuc ish 18cen(D18Z1x3) 47,XY,+i(18)(p10) isochromosome 18p isochromosome 18q
	Q91.4	Trisomy 13, meiotic nondisjunction	47,XX,+13 47,XY,+13
Patau's syndrome/Trisomy 13	Q91.4-Q91.7		
	Q91.5	Trisomy 13, mosaicism (mitotic nondisjunction)	46,XX/47,XX,+13 47,XY,+13/46,XY 46,XX[4}/47,XX,+13[6]
	Q91.6	Trisomy 13, translocation	46,XX,der(13;14)(q10;q10).+13 Unbalanced Robertsonian translocation - trisomy 13 NOT **45**,XX,der(13;14)(q10q10) which is balanced
	Q91.7	Patau's syndrome, unspecified	nuc ish 13q14(RB1x3)
Other trisomies and partial trisomes of autosomes	Q92	Other trisomies and partial trisomies of the autosomes, NEC Includes: unbalanced translocations and insertions Excludes: trisomies of chromosomes 13, 18, 21 (Q90-Q91)	
	Q92.0	Whole chromosome trisomy, meiotic nondisjunction	47,XY,+9 47,XX,+** Trisomy 22
	Q92.1	Whole chromosome trisomy, mosaicism (mitotic nondisjunction)	Trisomy 16 mosaic 47,XX,+9/46,XX
	Q92.2	Major partial trisomy Whole arm or more duplicated.	
	Q92.3	Minor partial trisomy Less than whole arm duplicated	46,XX,add(4)(q34) 46,XY,dup(15)(q25.2q25.3).ish dup(15)(535P8++) Trisomy 8p Use for all duplications including those detected by array
	Q92.4	Duplications seen only at prometaphase	Not yet of use
	Q92.5	Duplications with other complex rearrangements	Now selected as code for all unbalanced translocations 46,XX,der(3)t(1;3)(q25;p26) 46,XY,der(1)t(1;8)(q44;p11.21).ish der(1)(160H23-,52M11+)
	Q92.6	Extra marker chromosomes	47,XX,+mar(15) de novo 46,XY,+mar der(22) 47,XY,+mar pat.ish dic(22)
	Q92.7	Triploidy and polyploidy	69,XXX 69,XYY 92,XXYY
	Q92.8	Other specified trisomies and partial trisomies of autosomes	47,XY,i(12)(p10) / 46,XY
	Q92.9	Trisomy and partial trisomy of autosomes, unspecified	
Monosomies and deletions from the autosomes	Q93	Monosomies and deletions from the autosomes, NEC	46,XX,del(8)(p)
	Q93.0	Whole chromosome monosomy, meiotic nondisjunction	Only at miscarriage
	Q93.1	Whole chromosome monosomy, mosaicism (mitotic nondisjunction)	46,XX / 45,XX,-22
	Q93.2	Chromosome replaced with ring or dicentric	46,XX,r(10)(p15;q26.3)
Wolff-Hirschorn syndrome	Q93.3	Deletion of short arm of chromosome 4 Wolff-Hirschorn syndrome	46,XY,del(4)(p10) 46,XX,del(4)(p…)

Monitoring Group (MG) Condition	BPA Code	BPA Condition	Examples of written karyotypes and comments
Cri-du-chat syndrome	Q93.4	Deletion of short arm of chromosome 5 Cri-du-chat syndrome	46,XY,del(5)(p15.1) 46,XX,del(5)(p….)
	Q93.5	Other deletions of part of a chromosome Deletion of long arm of chromosome 13 Deletion of long or short arm of chromosome 18 [18p- or 18q syndrome]	46,XX,del(18)(p11.21)de novo 46,XX,ish del(15)(q11.2q11.2)(D15S13-) 46,XY,del(1)(p36.3) 46,XY,del(15)(q11.1->q12) Use for all deletions including those detected by array
	Q93.50	Deletion of long arm of chromosome 21 Anti-mongolism syndrome	46,XX,del(21)(q11-qter)
	Q93.6	Deletions seen only at prometaphase	Now selected as code for syndromic microdeletions with a specific ICD-10 syndrome code. Use syndrome code *and* Q93.6 if confirmed microdeletion. 46,XX,del(22)(q11.2q11.1).ish
	Q93.7	Deletions with other complex rearrangements	
	Q93.8	Other deletions from the autosomes	
	Q93.9	Deletion from autosomes, unspecified	
Turner's syndrome	Q96	Turner's syndrome Excludes: Noonan's syndrome (Q87.14)	
	Q96.0	Karyotype 45,X	Turner's syndrome
	Q96.1	Karyotype 46,X iso (Xq)	46,X,ipsudic(X)(p22.2)
	Q96.2	Karyotype 46,X with abnormal sex chromosome, except iso (Xq)	46,XX,del(X)(p22.31) 46,X,r(X)(p21q21) 46,X,del(X)(q21.1)
	Q96.3	Mosaicism, 45,X/46,XX or XY	45,X[10]/46,XY[12] Mosaic Turner's syndrome
	Q96.4	Mosaicism, 45,X/other cell line(s) with abnormal sex chromosome	45,X/47,XXX 45,X/47,XXY 45,X/46,XYY 45,X/46,XX/47,XXX 45,X[4]/46,XY[6]/47,XXY[2] mos 45,X/46,X,r(X)(p21q21)
	Q96.8	Other variants of Turner's syndrome	Not sure there is a use for this code
	Q96.9	Turner's syndrome, unspecified	PM findings highly suggestive of Turner's syndrome
	Q97.0	Karyotype 47,XXX	
	Q97.1	Female with more than three X chromosomes	48,XXXX 49,XXXXX Pentasomy X
	Q97.2	Mosaicism, lines with various numbers of X chromosomes	45,X / 46,XX / 47,XXX
	Q97.3	Female with 46, XY karyotype Excludes: Drash syndrome (N07)	
	Q97.8	Other specified sex chromosome abnormalities, female phenotype	46,X,dir dup(X)(q24q26.1)mat
	Q97.9	Sex chromosome abnormality, female phenotype, unspecified	
	Q98.0	Klinefelter's syndrome karyotype 47,XXY	
Klinefelter's syndrome	Q98.0-Q98.4		
	Q98.1	Klinefelter's syndrome, male with more than two X chromosomes	48,XXXY 49,XXXXY 49,XXXYY
	Q98.2	Klinefelter's syndrome, male with 46,XX karyotype	*Do not use*
	Q98.3	Other male with 46,XX karyotype	46,XX +SRY 46,XX in phenotypic male
	Q98.4	Klinefelter's syndrome, unspecified	*Do not use?*
	Q98.5	Karyotype 47,XYY	
	Q98.6	Male with structurally abnormal sex chromosome	46,X,del(Y)(q11.21) 46,X,der(Y)(p** or q**)
	Q98.7	Male with sex chromosome mosaicism	47,XYY / 46,XY 47,XXY / 46,XX 46,XY / 47,XYY / 45, X
	Q98.8	Other specified sex chromosome abnormalities, male phenotype	
	Q98.9	Sex chromosome abnormality, male phenotype, unspecified	
	Q99.0	Chimera 46,XX/46,XY Chimera 46,XX/46,XY true hermaphrodite	

Monitoring Group (MG) Condition	BPA Code	BPA Condition	Examples of written karyotypes and comments
	Q99.1	46,XX true hermaphrodite 46,XX with streak gonads 46,XY with streak gonads Pure gonal dysgenesis	
	Q99.2	Fragile X chromosome Fragile X syndrome	
	Q99.8	Other specified chromosome abnormalities	
	Q99.9	Chromosomal abnormality, unspecified	