

Aneuploidy, or abnormal chromosome copy number, is the leading cause of congenital birth defects and mental retardation.<sup>1</sup> Aneuploidy is technically possible in any chromosome, but only some occur with enough frequency to be named and characterized. Patau Syndrome (trisomy 13), Edward’s Syndrome (trisomy 18), and Down Syndrome (trisomy 21) are the most common congenital aneuploidies, and all result in severe intellectual impairment and various physical abnormalities, including hypotonia, spinal cord/skull deformation, and heart defects.<sup>1</sup>

Sex chromosome aneuploidy is another form of copy number variation. In Klinefelter’s Syndrome (XXY) males have an extra X chromosome, and generally experience language delays.<sup>2</sup> The female equivalent of Klinefelter’s, XXX Syndrome, comes with similar intellectual setbacks.<sup>2</sup> Infants with Turner’s Syndrome lack an X chromosome (X0 genotype) and have deficits in spatial ability.<sup>2</sup> Most sex chromosome aneuploidies also result in abnormal phenotypes, including incomplete development of secondary sex characteristics and little to no gamete production.<sup>2</sup>

Empire Genomics’ chromosome control probes can be used to detect aneuploidy. They test for the presence of whole chromosomes by hybridizing to the centromere. A complete chromosome count can then be made, with absent or extra signals indicating aneuploidy. Each probe comes in a set of 10 and normally ship within 7-10 business days.

PRODUCT	REGION	SKU
Chromosome 1	1p11-1q12	CHR01-10-GR
Chromosome 2	2q11.1	CHR02-10-GR
Chromosome 3	3p11.1	CHR03-10-GR
Chromosome 4	4p11	CHR04-10-GR
Chromosome 5	5p12-p11	CHR05-10-GR
Chromosome 6	6p11-q11	CHR06-10-GR
Chromosome 7	7q11.1	CHR07-10-GR
Chromosome 8	8p11-q11	CHR08-10-GR
Chromosome 9	9q21.3	CHR09-10-GR
Chromosome 10	10p-q11	CHR10-10-GR
Chromosome 11	11p11.11-q11	CHR11-10-GR
Chromosome 12	12p11-q11	CHR12-10-GR

PRODUCT	REGION	SKU
Chromosome 13	13q21.31	CHR13-10-GR
Chromosome 14	14q11.2	CHR14-10-GR
Chromosome 15	15q11.2	CHR15-10-GR
Chromosome 16	16p11.1	CHR16-10-GR
Chromosome 17	17p11.1	CHR17-10-GR
Chromosome 18	18p11.1-q11.1	CHR18-10-GR
Chromosome 19	19p13.11	CHR19-10-GR
Chromosome 20	20p11.21	CHR20-10-GR
Chromosome 21	21q21.1	CHR21-10-GR
Chromosome 22	22q13.32	CHR22-10-GR
Chromosome X	Xq11.1	CHRX-10-GR
Chromosome Y	Yp11.1 - Yq11.1	CHRY-10-GR

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1. Witters G., et al. (2011) Facts, views & vision in ObGyn 3.1: 15.  
2. Printzlaw F., et al. (2017) Jour neuro research 95.1-2: 311-319.