Sequencing-Based Karyotyping - User guide

What this is
We provide a genome-wide karyotype using whole-genome sequencing to detect the sex of the cell line, whole-chromosome aneuploidies, segmental gains/losses and copy number changes as low as 0.5M base. We use DNA sequencing to generate copy-number profiles across all chromosomes and summarize the result as a karyotype call for each sample.

How it works
Your DNA (or cell lysate) is converted to sequencing libraries using whole-genome amplification workflow, then sequenced. Our pipeline computes copy-number across the genome and reports the inferred karyotype, with plots and a brief interpretation.

What to provide
PCR grade genomic DNA sample. If you have performed PCR and Sanger sequencing for your gene/site of interest, it will be suitable for analysis. If you have not done that quality control, provide a concentration. Use the table below to provide required information.

1. Option A: Purified genomic DNA: concentration ≥ 10 ng/µL, volume ≥ 10 µL (in nuclease-free water).
2. Option B: QuickExtract lysate (e.g. use Lucigen QE09050): volume ≥ 20 µL.
3. Option C: cell pellet or picked colony in PCR vial >1000 cells

Turnaround time
20 business days

What you receive
1. A per-sample PDF with genome-wide copy-number plots and the summarized karyotype call (e.g., whole-chromosome gains/losses, segmental changes, and large CNVs greater than 0.5Mb with chromosomal coordinates).
2. Optional: raw data (FASTQ/BAM) upon request.

Sequencing-Based Karyotyping vs. Traditional G-Banding

	Our Service (Sequencing-based karyotyping)	**Traditional G-banded karyotyping**
Prep steps	Purified DNA or QuickExtract lysate - No added work for growing cells or DNA extraction beyond the one used to prepare for PCR and Sanger sequencing	Whole liquid-filled flask of live cells
Risk of sample failure	Negligible if instructions are followed	Sensitive to transport, contamination, and cell growth/culture issues
Price (per sample)	$50	~$500

Limitations

1. Cannot detect balanced rearrangements, e.g., translocations/inversions without copy number change.
2. No metaphase spread images — evaluate balanced translocations with G-banding/FISH.

This service is supported by the Molecular Genetics and DIsease Modeling Core (NYNORC P30, NIDDK) and the Columbia Stem Cell Core.

Please provide the following Table with information along with your samples:

Sample ID	Submitter name/group	Tube label	Species	DNA isolation method or cell pellet?	PCR/ Genotyping performed?	Concentration	Contact email/uni

EXAMPLE

Cell Line Karyotyping Report

Project Summary

Sample ID	Species	Ploidy	Karyotype
Example 1	Homo sapiens	Euploid	46,XY
Example 2	Homo sapiens	Aneuploid	46,XY,dup(1)(q32.1)
Example 3	Homo sapiens	Aneuploid	47,XX,+1,del(14)(q32.12-q32.33)

Copy Number Variation Graph

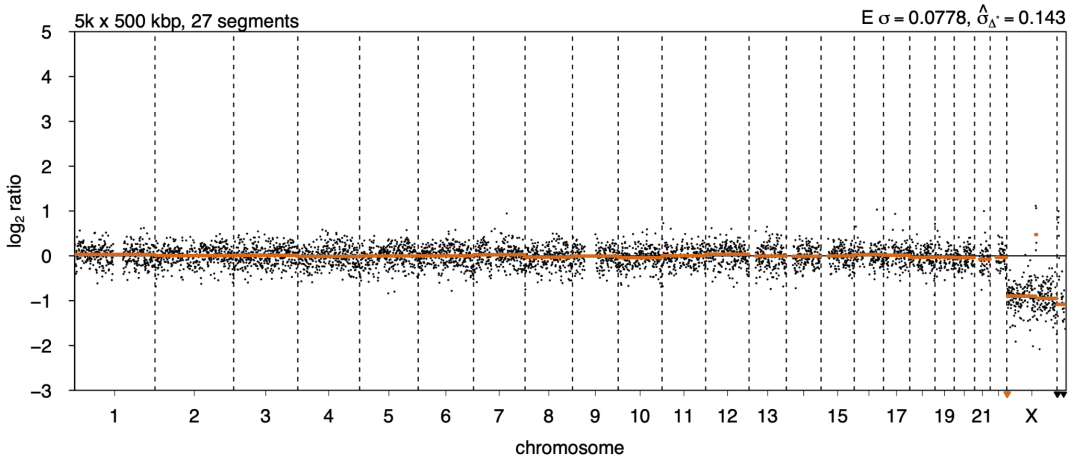
*Log_2 ratio = log_2 (sample copy number/reference copy number)

0	2 copies
> 0.5	Gain of 1 or more copies
-1	1 copy
< -2	0 copy/nullisomy

Example 1

Reference genome: hg19

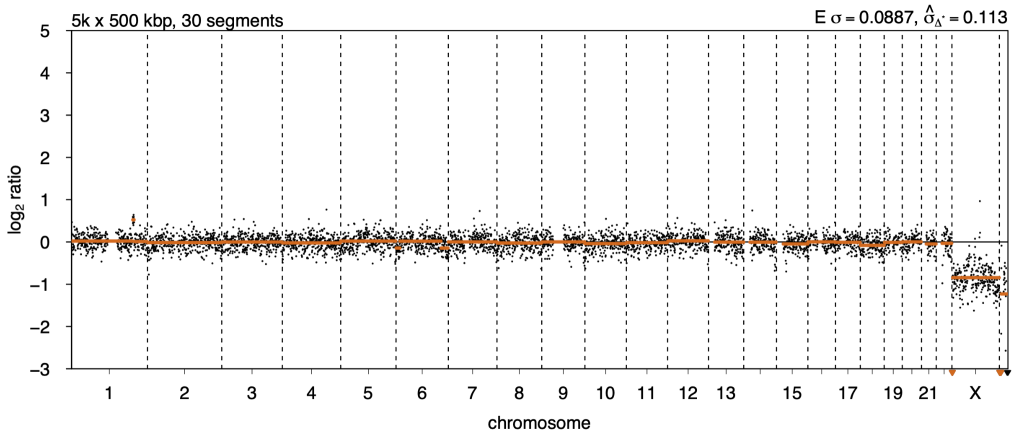
No chromosomal aberrations were found compared to the reference dataset.



Example 2

Reference genome: hg19

Chrom	Start Position	End Position	Size (Mb)	Log_2 Ratio	Copy Number	Status
1	201500001	206000000	4.5	0.524	2.876	Gain



Example 3

Reference genome: hg19

Chrom	Start Position	End Position	Size (Mb)	Log_2 Ratio	Copy Number	Status
1	1000001	248500000	247.5	0.412	2.661	Gain
14	94000001	106000000	12.0	-0.776	1.168	Loss