The American Journal of Human Genetics, Volume *105*

Supplemental Data

Validation Studies for Single Circulating Trophoblast Genetic Testing as a Form of Noninvasive Prenatal Diagnosis

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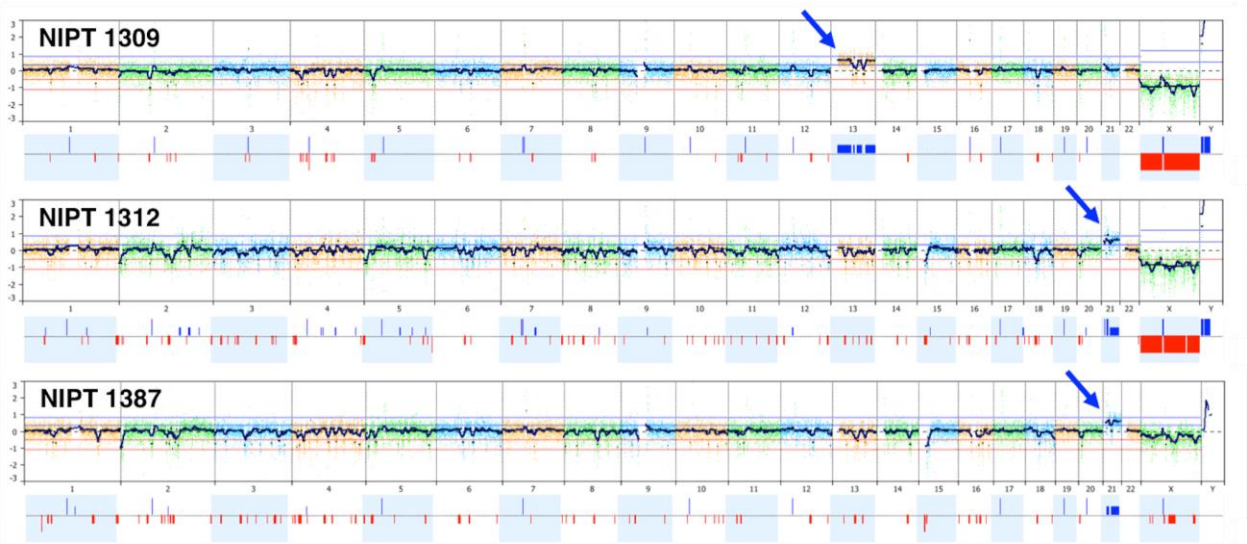

Figure S1. S-phase cells for aneuploidy calling. This figure includes three examples of single trophoblasts each showing a pattern indicating the cell being in S-phase, but that are nevertheless still useful for calling a trisomic event. These whole genome plots were generated by comparing to a normal female reference: the two top cells were obtained from a male fetus, while the third cell was recovered from a female pregnancy.

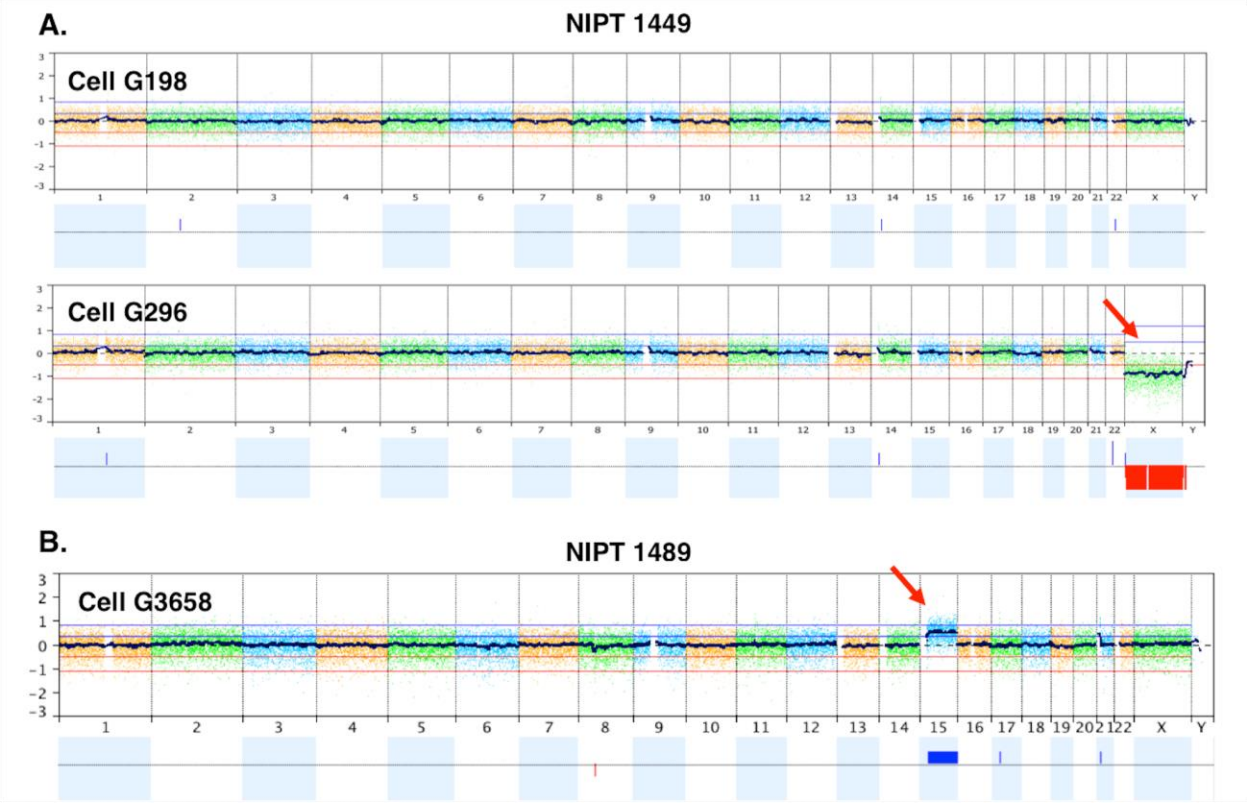

Figure S2. Confined placental mosaicism. The two plots in panel A show two of the seven scorable trophoblasts found for case NIPT1449, illustrating the mosaic 45,X/46,XX result that was seen for this sample. Cell G198 shows a normal 46,XX complement compared to a normal female reference, while cell G296 shows a clear loss of one copy of X. Panel B shows the trophoblast doublet that was analyzed for NIPT1489, in which a clear trisomy 15 was seen.

Table S2. Overview of samples from twin pregnancies

Case	GA	Fetal sex	Cells/mL	Total # of cells sequenced	# of high-quality cells	# of cells with aneuploidy data only	
NIPT1329	12w3d	M + F	0.33	7	4	3	Cells from both fetuses
NIPT1334	13w3d	M + M	0.40	6	4	0	*(+2 poor quality cells)*
NIPT1432	19w	M + F	0.11	1	1	0	Data for only 1 fetus
NIPT1441	13w6d	M + F	0.77	10	3	7	Data for only 1 fetus (only female cells were recovered)
NIPT1452	13w2d	F + F	0.65	8	2	2	*(+4 poor quality cell)*
NIPT1482	13w	M + F	0.25	5	3	1	*(+1 poor quality cell)* Cells from both fetuses
NIPT1495	12w	M + M	0.33	9	5	1	*(+1 poor quality cell; +2 cells of maternal origin)*
NIPT1496	12w2d	M + F	0.40	10	7	2	*(+1 poor quality cell)* Cells from both fetuses

Table S3. NGS results for all singleton cases collected ≥15 weeks gestation

NGS result	Study 1 (8 cases)	Study 2 (9 cases)
≥ 2 high-quality cells	1 case (12.5%)	1 case (11.1%)
1 high-quality cell	3 cases (37.5%)	6 cases (66.7%)
Aneuploidy data only	2 cases (25.0%)	1 case (11.1%)
No usable data	2 cases (25.0%)	1 case (11.1%)