

## Next-generation counseling: noninvasive prenatal screening results disclosure and management

### Supplementary Material

Aishwarya ARJUNAN, MS, MPH, CGC<sup>1</sup>; Rotem BEN-SHACHAR, PhD<sup>1</sup>; Jamie KOSTIALIK, MS, CGC<sup>1</sup>; Katherine JOHANSEN TABER, PhD<sup>1</sup>; Gabriel A. LAZARIN, MS, CGC<sup>1</sup>; Elizabeth DENNE, MS, CGC<sup>1</sup>; Dale MUZZEY, PhD<sup>1</sup>; Carrie HAVERTY, MS, CGC<sup>1</sup>

<sup>1</sup>Counsyl, 180 Kimball Way, South San Francisco, CA 94080, USA

Conflict of Interest/Disclosure: All authors are employees of Counsyl.

### Supplementary Table I. Patient Ethnicities of Diverse Patient Cohort

<b>Ethnicity</b>	<b>Tested</b>	<b>% (95% CI)</b>
African/African-American	4,461	6.7 (6.4, 7.0)
Ashkenazi Jewish	852	1.3 (1.1, 1.4)
East Asian	2,290	3.4 (3.2, 3.7)
French Canadian/Cajun	118	0.18 (0.1, 0.2)
Finnish	11	0.02 (.007, .04)
Hispanic	6,606	10.0 (9.6, 10.3)
Middle Eastern	803	1.2 (1.1, 1.3)
Mixed/Other Caucasian	23,046	34.7 (34.7, 35.2)
Native American	225	0.3 (0.3, 0.4)
Northern European	7,213	10.9 (10.5, 11.2)
Pacific Islander	166	0.2 (0.2, 0.3)
South Asian	1,986	3.0 (2.8, 3.2)
Southeast Asian	1,412	2.1 (2.0, 2.2)
Southern European	938	1.4 (1.3, 1.6)
Unknown/Not reported	16,348	24.6 (24.1, 25.1)

### Supplementary Table II: Screen Positive Results

<b>Condition Screened</b>	<b>Screen Positive Results</b>
Trisomy 13*	110
Trisomy 18*	176
Trisomy 21*	458
Monosomy 13	22
Monosomy 18*	12
Monosomy 21	9
Monosomy X	253
XXX	56
XXY	53
XYY	28
del1p36	7
del5p	8
del4p	6
del15q11.2	12
del22q	8
Total	1,218 <sup>†</sup>

\*Includes both aneuploidy suspected and aneuploidy detected results

<sup>†</sup> Some tests were positive for multiple conditions: 1180 tests were positive for one condition, 16 tests were positive for two conditions and 2 tests were positive for three conditions.

**Supplementary Table III: Factors affecting genetic counseling election**

	<b>Positive Test Results Delivered by Provider</b>	<b>Positive Test Result Delivered by Provider and Elected Genetic Counseling</b>	<b>Negative Test Results</b>	<b>Negative Test Result and Elected Genetic Counseling</b>
AMA, first pregnancy	137	19 (13.8%)	7,427	746 (10.0%)
AMA, not first pregnancy	517	75 (14.5%)	21,562	1,765 (8.2%)
Abnormal U/S, central nervous system	33	3 (9.1%)	504	36 (7.1%)
Abnormal U/S, other	113	14 (12.4%)	2,180	144 (6.6%)
Chromosome abnormality suspected in fetus	57	8 (14.0%)	785	68 (8.6%)
Other high-risk pregnancy	55	11 (22.0%)	3,027	262 (8.7%)
Family history	10	1 (10.0%)	860	89 (10.3%)
History of chromosomal abnormality in previous pregnancy	11	3 (27.3%)	831	86 (10.3%)
Abnormal maternal serum screen	48	8 (16.7%)	1,413	100 (7.1%)
Twin pregnancy	15	1 (6.7%)	1,388	134 (9.7%)
Tried IVF	58	11 (19.0%)	3,240	243 (7.5%)
Provider delivered results	1,166	194 (16.6%)	2,172	40 (1.8%)
Positive test results	1,166	194 (16.6%)	0	0 (0%)

AMA: Advanced Maternal Age

U/S: Ultrasound

IVF: In-vitro fertilization