

NIPT COMPARISON CHART

Illumina	MaterniT21	MaterniT21 PLUS	MaterniT GENOME	Condition	Chromosome Condition	Prevalence in general population	Affects
x	x	x	x	Down syndrome	Trisomy 21 (extra 21st chromosome)	1 in 800	Heart defects, mental delay, other
x	x	x	x	Edward syndrome	Trisomy 18 (extra 18th chromosome)	1 in 5000	Variable but typically heart defects, marked mental delay, other. Neonatal death
x	x	x	x	Patau syndrome	Trisomy 13 (extra 13th chromosome)	1 in 16000	Multiple severe abnormalities including heart defects, cleft lip/ palate, kidney abnormalities, other. Foetal or neonatal death.
x	x	x	x	Turner syndrome	X0 (missing X chromosome)	1 in 2500 (girls)	Short stature, may have heart defects, kidney, skeletal abnormalities, webbed neck, infertility, other. May die before birth Normal intelligence
x	x	x	x	Sex chromosome abnormalities	XXX, XYY, XYY, XXY (Klinefelter syndrome)	1 in 500 Klinefelter affects 1 in 1000 males	Usually mild to few effects Klinefelter with infertility, may have learning disabilities
		x	x	DiGeorge syndrome	22q11-	1 in 4,000	Heart defects, cleft palate, immune deficiency, learning disabilities, delay, other
		x	x	Cri-du-chat syndrome	5p-	1 in 20,000 to 50,000	Severe developmental delay, microcephaly, hypotonia, heart defects
		x	x	Angelman syndrome Prader-Willi Syndrome	15q-	1 in 10,000 to 30,000	Developmental delay, other
		x	x	1p36 deletion syndrome	1p-	1 in 10,000	Developmental delay, brain abnormalities, heart defects, other
		x	x	Jacobsen syndrome	11q	1 in 100,000	Developmental delay, cognitive impairment, bleeding disorder, other
		x	x	Langer-Giedion syndrome	8q	Rare	Learning difficulties, short stature, skeletal abnormalities, other
		x	x	Wolf-Hirschhorn syndrome	4p	1 in 50,000	Developmental and growth delay, intellectual disability, hypotonia, seizures, other
		x	x	Trisomy 16	Trisomy 16 (extra 16th chromosome)	1 in 100 pregnancies 1 in 50,000 births	Multiple abnormalities leading to abortion or early foetal death
		x	x	Trisomy 22	Trisomy 22 (extra 22nd chromosome)	1 in 200 pregnancies 1 in 40,000 births	Multiple abnormalities leading to abortion or early foetal death
			x		Whole chromosome abnormalities, all chromosomes		
			x		Partial chromosome abnormalities, all chromosomes		
3 -5 working days	5-7 working days	5-7 working days	5-7 working days	Turnaround time	From blood draw		

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10	10	10	10	Draw week			
	> 1M	> 1M	> 80,000	Number of tests done			
< 2%	< 1%	< 1%	~ 2%	Failure rate	Non-reportable result		
> 16,000	>40,000	> 40,000	-	Twin pregnancies			
-	x	x	-	Triplets or higher multiple			
x	x	x	x	Egg donor/IVF			
	x	x	x	Report format	Positive/Negative		
x				Report format	Risk evaluation		