SUPPLEMENTAL METHODS

Sample collection and DNA extraction

Pregnant Ashkenazi Jewish (AJ) couples, carrying mutation/s in the *GBA* gene, were recruited at the Shaare Zedek Medical Center (SZMC) Gaucher Clinic. Peripheral blood samples were collected from each couple, relevant mutation carrier family members, 8 unrelated AJ *GBA* N370S homozygotes, and 3 unrelated AJ *GBA* N370S heterozygote duos. Genomic DNA was then prepared from all samples using the FlexiGene DNA kit (QIAGEN) according to the manufacturer's protocol. For pregnant female indices, plasma was separated from peripheral blood by centrifugation at 1,900 x *g* for 10 minutes at 4°C. The plasma supernatant was then recentrifuged at 16,000 x *g* for 10 minutes at 4°C and 3ml of the resulting supernatant was used for cell-free DNA extraction with the QIAamp Circulating Nucleic Acid kit (QIAGEN) according to the manufacturer's protocol. The maternal plasma DNA extracts were then pre-amplified, in duplicate, with the SurePlex Amplification System (Illumina) ahead of downstream processing. All familial mutations in *GBA* were Sanger sequence verified prior to commencement of the study. Ethical approval for the study, including usage of materials from human subjects, was obtained from the local institutional review board and written informed consent was obtained from all study participants.

Next generation sequencing (NGS) of GBA-flanking single nucleotide polymorphisms (SNPs)

Two TruSeq Custom Amplicon panels were designed with DesignStudio software (Illumina) to amplify and sequence *GBA*-flanking SNPs in all samples. The smaller panel sequenced 490 SNPs and the larger panel sequenced 5,000 SNPs. Indexed next generation sequencing libraries were prepared and normalized according to the manufacturer's protocol (Illumina) followed by 2x150bp pair-end sequencing on a MiSeq (small panel) or NextSeq 500 (large panel) instrument (Illumina) to a mean depth

of at least 500x or 3800x for genomic and plasma DNA samples, respectively. After sequencing runs, the data were aligned to target sequences on the human reference genome (hg19) using MiSeq Reporter software (Illumina) for the small panel or the TruSeq Amplicon v1.1 app on BaseSpace (https://basespace.illumina.com/) for the large panel. Genotyping data was extracted from each alignment using the SAMtools mpileup program to yield sample-specific SNP genotype profiles and then the SNPs were annotated by snpEff with dbSNP138 (small panel) or dbSNP141 (large panel). These profiles were then combined into single family-specific .csv files using in-house software so as to facilitate familial and fetal linkage analysis (see below). Prior to linkage analysis, non-*GBA* flanking SNP calls and SNP calls on heavily self-chained genomic segments were removed. Genomic DNA SNP genotype calls were categorized into one of 3 distinct classifications based on the percentage of non-reference genome allele (B allele) sequencing reads at each locus: homozygote reference allele (AA; 0%-20% B allele reads); homozygote non-reference allele (BB; 80%-100% B allele reads); or heterozygote (AB; 30%-70% B allele reads). Any loci that did not meet these classification criteria were excluded from further downstream analysis. As a rule, parental haplotypes were constructed with SNPs for which the parent was heterozygous and at least one of his/her first degree relatives was homozygous.

Construction of consensus AJ N370S and familial haplotypes

The initial consensus AJ N370S *GBA*-flanking haplotype was constructed by performing homozygosity mapping with custom SNP small panel NGS datasets from 7 unrelated AJ N370S homozygotes (14 N370S chromosomes). Subsequently, 6 more AJ N370S haplotypes were derived from linkage analysis on SNP NGS datasets from 6 unrelated AJ N370S mutation carrier duos. Each linkage-based N370S haplotype was then crossed with the consensus sequence derived from homozygosity mapping to identify inconsistencies. These sequence discrepancies were then used to mark consensus AJ N370S founder haplotype cut-offs (based on 20 N370S chromosomes, altogether, after the completion of all data

intersections). The larger consensus AJ N370S GBA-flanking haplotype was constructed by performing homozygosity mapping with custom SNP large panel NGS datasets from 8 unrelated AJ N370S homozygotes (16 N370S chromosomes). Subsequently, 12 more AJ N370S haplotypes were derived from linkage analysis on SNP NGS datasets from 12 unrelated AJ N370S mutation carrier duos. The final consensus AJ N370S founder haplotype cut-offs (based on 28 N370S chromosomes, altogether, after the completion of all data intersections) were then set as described above regarding the initial consensus haplotype construct.

Identification of fetal alleles in maternal plasma DNA

In order to construct credible small fetal haplotypes (composed of <5 SNPs) with the small SNP sequencing panel, plasma DNA samples were sequenced in duplicate at high depth (>3,000x mean coverage) so as to augment statistical confidence in each individual fetal SNP genotype call. In all, four different combinations of parental SNP genotypes were analyzed in plasma DNA: A) Error rate informative (father and mother [of the fetus] both homozygote "AA"); B) Dosage informative (father and mother homozygote for opposite alleles); C) Paternal haplotype informative (father heterozygote and mother homozygote); and D) Maternal haplotype informative SNPs (mother heterozygote and father homozygote). Error rate informative SNPs measured the sequencing error rate in plasma DNA samples by assessing the appearance of biologically impossible SNP reads. At >1000x read depth, error rates of 0.6% +/- 0.6% were measured in plasma DNA samples. Dosage informative SNPs (denoted heretofore as "SNP I") measured the paternal portion of fetal plasma DNA by determining the fraction of paternal alleles per maternal alleles. These SNPs also confirmed the presence of fetal DNA in maternal plasma. Paternal haplotype informative SNPs (denoted heretofore as "SNP II") feature a unique nucleotide in the fetus' father that is not present in the maternal genotype. When identified in maternal plasma DNA, the paternal unique allele is expected to comprise the same fraction as those of paternal alleles in dosage

informative SNPs. In general, the paternal haplotype of the fetus was deduced wherever the father's unique SNP II allele was identified in one of 2 plasma DNA replicates (at a SNP position with >1000x sequencing depth) with relatively high frequency (>2σ from the mean sequencing error rate as determined from error rate informative SNPs) in maternal plasma DNA. The computed sensitivity/specificity scores for this method are provided as a function of the number of unique paternal SNPs identified in the fetus (see Supplemental Table 1).

For plasma DNA samples with high fetal dosage (>30% paternal fetal fraction), the paternal haplotype in the fetus was also deduced from non-unique SNP II alleles (with >500x coverage) for which there were no discrepancies between replicate fetal haplotype calls. The computed sensitivity/specificity scores for this method are provided as a function of the number of non-unique paternal SNPs identified in the fetus (see Supplemental Table 2). Maternal haplotype informative SNPs (denoted heretofore as "SNP III") were used to determine the maternal haplotype in the fetus at >1000x sequencing coverage. These SNPs indicated a heterozygous fetal genotype when allele-allele ratios were balanced, and a homozygous fetal genotype when these ratios were imbalanced by a number >3σ from the mean sequencing error rate (as determined from error rate informative SNPs). Depending on the father's homozygous allele, the maternal fetal allele was deduced based on the presence or absence of skewing (<50% non-reference nucleotide skewed representation if the father was homozygote A [for the reference nucleotide]; >50% non-reference nucleotide skewed if the father was homozygote B [for the non-reference nucleotide]) in maternal heterozygous SNP III loci on both plasma DNA replicates. The computed sensitivity/specificity scores for this method are provided as a function of the number of maternal haplotyped SNPs identified in the fetus (see Supplemental Table 3). All parental SNP combinations that did not fall within the above guidelines were not utilized in this study.

In order to construct large fetal haplotypes (composed of >5 SNPs) with the large SNP sequencing panel, plasma DNA samples were analyzed as above with the following modifications. Error rate informative

SNPs indicated a 1% error rate at read depths exceeding 100x. Accordingly, paternal haplotype informative and maternal haplotype informative SNPs were assessed from a minimum read depth of 100 whereupon only skewing exceeding 1% B-allele frequency in plasma DNA with respect to maternal DNA (at a particular locus) was considered significant enough for incorporation into the fetal haplotype. This filter was applied so as to reduce genotyping errors emerging from either sequencing error and/or off-target sequence contamination.

Ultimately, fetal diagnosis was achieved after comparing the paternal and maternal cell-free fetal DNA (cffDNA) haplotypes with family-based and/or N370S consensus or near consensus haplotypes as relevant. Altogether, the entire noninvasive NGS-based prenatal test, from blood sample processing to fetal diagnosis, was completed in 5 work days. In addition, all diagnoses were confirmed by post-natal genetic testing. For family 1, allelic inheritance of the N370S mutation was further confirmed by postnatal linkage analysis with short tandem repeat (STR) markers.

Supplemental Table 1. Simulated sensitivity/specificity for unique paternal allele diagnosis

No. SNPs in fetal haplotype	Sensitivity/Specificity[A]
1	94.97%
2	99.72%
3	99.96%
4	99.97%
5	100.00%
6	100.00%
7	100.00%
8	100.00%
9	100.00%
10	100.00%

A. The formula for these calculations was as follows: $[1-([(0.5)(er)]+[(0.5)(er)])^n]$ where "n" represents the number of SNPs in the fetal haplotype and "er" represents the chance (which is 5%) of unique paternal allele detection at 2σ from the sequencing error rate as determined from error rate informative SNP sequences (see Supplemental Methods). For 1 to 4 SNP haplotypes, a 0.03% correction was applied to account for the sex-specific male recombination rate in the +/- 250kb genomic region surrounding *GBA* according to reference (20) but if longer haplotypes do not flank the mutation, this correction should continue to be applied.

Supplemental Table 2. Simulated sensitivity/specificity for non-unique paternal allele diagnosis

No. SNPs in fetal haplotype	Sensitivity/Specificity[A]
1	77.41%
2	94.88%
3	98.82%
4	99.71%
5	99.94%
6	99.99%
7	100.00%
8	100.00%
9	100.00%
10	100.00%

A. The formula for these calculations was as follows: $[1-([(0.5)(1-er)]^2)^n]$ where "n" represents the number of SNPs in the fetal haplotype and "er" represents the chance (which is 5%) of unique paternal allele detection at 2σ from the sequencing error rate as determined from error rate informative SNP sequences (see Supplemental Methods). For 1 to 4 SNP haplotypes, a 0.03% correction was applied to account for the sex-specific male recombination rate in the +/- 250kb genomic region surrounding *GBA* according to reference (20) but if longer haplotypes do not flank the mutation, this correction should continue to be applied.

Supplemental Table 3. Simulated sensitivity/specificity for maternal allele diagnosis

No. SNPs in fetal haplotype	Sensitivity/Specificity[A]
1	74.93%
2	93.68%
3	98.37%
4	99.54%
5	99.90%
6	99.98%
7	99.99%
8	100.00%
9	100.00%
10	100.00%

A. The formula for these calculations was as follows: $[1-[(0.5)^2]^n]$ where "n" represents the number of SNPs in the fetal haplotype. For 1 to 4 SNP haplotypes, a 0.07% correction was applied to account for the sex-specific female recombination rate in the +/- 250kb *GBA* region according to reference (20) but if longer haplotypes do not flank the mutation, this correction should continue to be applied.

Supplemental Table 4. Parental family-based haplotype information

Family	Paternal familial haplotype data				Maternal familial haplotype data			
	Paternal genotype	Paternal family member used for linkage	Genotype of paternal family member	No. of SNPs in linked haplotype	Maternal genotype	Maternal family member used for linkage	Genotype of maternal family member	No. of SNPs in linked haplotype
1	N370S/WT	father	N370S/WT	3	N370S/WT	sister	N370S/WT	43
2	WT/WT	N/A	N/A	N/A	N370S/WT	mother	N370S/WT	11

Abbreviations: WT, wild type; N/A, not applicable

Supplemental Table 5. Consensus Ashkenazi Jewish N370S founder haplotype

Ch	Position (hg19)	dbSNP ID	REF	ALT	consensus AJ N370S haplotype[A]
1	155186381	rs148168407	G	.	A
1	155186551	rs147791517	C	.	A
1	155186648	rs372959451	T	.	A
1	155186669	rs184043244	G	.	A
1	155186716	rs375966138	A	.	A
1	155186729	rs28445596	T	.	A
1	155186742	rs1057941	G	.	A
1	155187761	rs185115702	C	.	A
1	155187786	rs149513274	G	.	A
1	155188130	rs138784147	A	.	A
1	155188811	rs115207152	A	.	A
1	155188856	rs183891209	G	.	A
1	155194980	rs2049805	T	.	A
1	155195044	rs6677756	A	G	B
1	155197462	rs2990245	C	.	A
1	155197553	rs201008360	G	.	A
1	155197554	rs3835732	T	.	A
1	155197556	rs377586853	G	.	A
1	155199308	rs181495640	G	.	A
1	155199315	rs12407919	C	.	A
1	155199356	rs186289485	G	.	A
1	155201235	rs1045253	G	.	A
1	155204239	*GBA* 3' UTR[B]	.	.	-
1	155214653	*GBA* 5' UTR[B]	.	.	-
1	155216184	rs1158151	T	.	A
1	155218365	rs734073	T	.	A
1	155218370	rs139803480	C	.	A
1	155218446	rs734074	A	.	A
1	155218476	rs192678801	C	.	A
1	155218487	rs185728978	G	.	A
1	155221837	rs2072647	C	.	A
1	155221844	rs2075567	C	.	A
1	155221862	rs2974918	C	.	A
1	155221890	rs180894227	C	.	A
1	155221927	rs186280028	C	.	A
1	155221942	rs190742122	C	.	A
1	155224091	rs2075566	C	.	A
1	155224123	rs2974915	A	.	A
1	155224144	rs200790772	C	.	A
1	155224154	rs377616889	A	.	A
1	155224301	rs2242577	C	.	A
1	155224393	rs12758281	G	.	A
1	155224404	rs368919969	G	.	A
1	155224417	rs140665962	C	.	A

1	155230131	rs1142287	C	.	A
1	155230134	rs138266025	C	.	A
1	155230144	rs143921371	T	.	A
1	155230157	rs201400281	G	.	A
1	155230187	rs371041272	G	.	A
1	155230212	rs149415891	C	.	A
1	155230222	rs373833483	G	.	A
1	155232071	rs111246834	C	.	A
1	155232074	rs1046188	C	.	A
1	155232093	rs375274930	G	.	A
1	155232189	rs368313864	A	.	A
1	155234821	rs1076556	A	.	A
1	155234827	rs199775545	A	.	A
1	155234895	rs12044394	T	.	A
1	155234911	rs12043655	A	.	A
1	155234914	rs148772014	G	.	A
1	155235493	rs1078699	T	.	A
1	155235535	rs760078	A	.	A
1	155235557	rs192046581	C	.	A
1	155239624	rs2361543	G	.	A
1	155239657	rs142820979	C	.	A
1	155241361	rs2236863	G	.	A
1	155241368	rs57336884	A	.	A
1	155241459	rs140225864	T	.	A
1	155247707	rs12749306	G	.	A
1	155247722	rs189401977	G	.	A
1	155248575	rs7549276	G	.	A
1	155248622	rs145609188	C	.	A
1	155248655	rs181989907	C	.	A
1	155252833	rs12049375	G	.	A
1	155253583	rs7520184	G	.	A
1	155255837	rs11264352	T	.	A
1	155255851	rs373919604	A	.	A
1	155255906	rs11264353	G	.	A
1	155257492	rs11264355	C	.	A
1	155257531	rs202118354	C	.	A
1	155257557	rs191318307	C	.	A
1	155257559	rs373694526	G	.	A
1	155260096	rs932972	G	.	A
1	155260153	rs35957800	G	.	A
1	155260348	rs374751259	G	.	A
1	155260349	rs8177994	G	.	A
1	155260350	rs1052177	A	.	A
1	155260360	rs368150636	G	.	A
1	155260382	rs61755431	C	.	A
1	155260383	rs1052176	G	.	A
1	155260402	rs140859641	G	.	A

1	155260415	rs370938255	C	.	A
1	155260434	rs370316462	C	.	A
1	155260443	rs190966689	C	.	A
1	155260448	rs375525211	C	.	A
1	155262603	rs8177984	T	.	A
1	155262613	rs4620533	C	.	A
1	155262638	rs8177983	T	.	A
1	155269776	rs3020781	A	.	A
1	155269780	rs8177964	G	.	A
1	155269830	rs8177963	A	.	A
1	155269863	rs377494351	G	.	A
1	155269864	rs369726754	G	.	A
1	155269865	rs199627687	C	.	A
1	155273869	rs4971072	G	A	B
1	155273917	rs141353488	C	.	A
1	155274960	rs12032720	G	.	A
1	155279482	rs2297480	T	.	A
1	155279550	rs144868148	C	.	A
1	155282829	rs11264359	A	.	A
1	155282884	rs34838448	T	.	A
1	155289545	rs11264361	T	.	A
1	155318308	rs11264363	G	C	B
1	155318313	rs138544252	T	.	A
1	155318353	rs368485885	T	.	A
1	155322142	rs12239114	G	.	A
1	155322147	rs148991117	T	.	A
1	155337338	rs11264367	T	.	A
1	155337373	rs34538089	A	.	A
1	155345043	rs5005770	A	.	A
1	155345056	rs374205358	T	.	A
1	155345077	rs73008959	G	.	A
1	155359817	rs72704164	A	.	A
1	155359820	rs188552875	A	.	A
1	155359830	rs11264369	T	C	B
1	155359880	rs6694257	C	.	A
1	155359905	rs374170158	G	.	A
1	155368203	rs7556102	C	A	B
1	155368210	rs56770523	T	.	A
1	155368233	rs375072511	G	.	A
1	155368288	rs73008989	C	.	A
1	155368294	rs201573542	T	.	A
1	155368311	rs183124840	T	.	A
1	155389688	rs10908465	C	.	A
1	155407083	rs146899245	G	.	A
1	155407096	rs12041534	C	.	A
1	155407111	rs114662639	A	.	A
1	155407180	rs149436342	A	.	A

1	155412930	rs11264372	A	.	A
1	155413304	rs1325908	C	A	B
1	155413331	rs151301522	T	.	A
1	155413360	rs72993428	C	.	A
1	155413374	rs376952825	C	.	A
1	155413386	rs79999785	T	.	A
1	155414729	rs10796943	T	.	A
1	155414768	rs12730906	C	.	A
1	155414806	rs375077645	G	.	A
1	155414813	rs12034526	T	.	A
1	155424065	rs11264375	T	.	A
1	155427062	rs6684889	T	A	B
1	155427148	rs1536255	C	.	A
1	155427688	rs10796945	G	.	A
1	155433942	rs12724079	T	C	B
1	155433971	rs140724364	G	.	A
1	155434017	rs114909331	A	.	A

Abbreviations: Ch, chromosome; REF, reference nucleotide (dbSNP Build 138); ALT, alternate (non-reference) nucleotide (dbSNP Build 138)

A. For the consensus AJ N370S haplotype: "A" = dbSNP reference nucleotide; "B" = dbSNP non-reference nucleotide

B. The region shaded in gray indicates *GBA* gene 5' and 3' locus boundaries

Supplemental Table 6. Identification of the paternal allele in the family 1 fetus (small panel)

Variant data							plasma DNA rep 1-1		plasma DNA rep 1-2		Haplotypes[E]			FAI[F]
Ch	Position (hg19)	dbSNP ID[A]	DFM (bps)	PGT[B]	MGT[B]	FL[C] (%)	RD (x)	BAF[D] (%)	RD (x)	BAF[D] (%)	PHiF	PFB N370S	N370S cons	DPAiF
1	155007293	rs4845404	198,341	**AB**	**BB**		7748	**64.4**	7692	98.6	A	-	-	-
1	155007302	rs4845405	198,332	**AB**	**BB**		7976	**66.4**	8000	99.7	A	-	-	-
1	155009258	rs11264298	196,376	**AB**	**BB**		1	0.0	1812	**67.7**	A	-	-	-
1	155028522	rs11264302	177,112	**AB**	**AA**	67.7	698	**0.3**	545	**0.2**	**A**	**B**	-	**WT**
1	155030557	rs11264303	175,077	**AB**	**AA**		7999	**0.9**	7999	**0.8**	**A**	**B**	-	**WT**
1	155205634	**N370S**	-	-	-		-	-	-	-	-	-	-	-

Abbreviations: Ch, chromosome; DFM, distance from mutation; PGT, paternal genotype; MGT, maternal genotype; FL, fetal load; rep, replicate plasma DNA sample, RD, sequencing read depth; BAF, B-allele frequency; PHiF, paternal haplotype in fetus; PFB N370S, paternal family-based N370S-linked haplotype; FAI, fetal allele identity; DPAiF, diagnosed paternal allele in fetus.

A. dbSNP ID or *GBA* mutation (red lettering)

B. For parental genotypes "AA" = homozygote dbSNP reference allele; "BB" = homozygote dbSNP non-reference allele; "AB" = heterozygote

C. Fetal load is 2x(mean paternal fetal fraction) as determined from SNP I and/or SNP II data (see Supplemental Methods)

D. B-allele frequency (BAF) is the % frequency of (B-allele reads)/(total read depth (RD)) at the indicated nucleotide position; bold BAF data was used to construct "PHiF"

E. The paternal fetal haplotype (PHiF) was determined from SNP II data (as described in Supplemental Methods); the paternal N370S-linked haplotype (PFB N370S) was determined from family-based linkage analysis; the N370S consensus haplotype (N370S cons) was derived according to Figure 2. An "-" indicates that no haplotype data was available at the given position. Bold alleles were used for diagnosis of the paternal allele in the fetus ("DPAiF").

F. Fetal allele identity (FAI) was determined by comparing the "PHiF" haplotype to the "PFB N370S" haplotype

Supplemental Table 7. Preliminary summary of noninvasive prenatal diagnosis with validation

plasma DNA sample information					Paternal haplotype in fetus			Maternal haplotype in fetus			Validation
Family	**Week gestation**	**Paternal genotype**	**Maternal genotype**	**Fetal load (%)**	**Diagnosis**	**Based on consensus N370S haplotype?**	**Based on familial haplotype?**	**Diagnosis**	**Based on consensus N370S haplotype?**	**Based on familial haplotype?**	
1	31	N370S/WT	N370S/WT	67.7	WT	NO[A]	YES	N370S	YES	YES	Postnatal
2	22	WT/WT	N370S/WT	5.7	N/A	N/A	N/A	N370S	YES	NO	Postnatal cord blood

Abbreviation: N/A, not applicable

A. Due to paternal homozygosity in consensus N370S haplotype region

Supplemental Table 8. Identification of the maternal allele in the family 1 fetus (small panel)

Variant data							plasma DNA rep 1-1		plasma DNA rep 1-2		Haplotypes[E]			FAI[F]
Ch	Position (hg19)	dbSNP ID[A]	DFM (bps)	PGT[B]	MGT[B]	FL[C] (%)	RD (x)	BAF[D] (%)	RD (x)	BAF[D] (%)	MHiF	MFB N370S	N370S cons	DMAiF
1	155047379	rs1001848	158,255	**AA**	**AB**		7876	13.1	3565	8.6	A	-	-	-
1	155068748	rs7365544	136,886	**AA**	**AB**		1604	30.9	1260	32.6	A	-	-	-
1	155079477	rs7367207	126,157	**AA**	**AB**		3082	44.4	4681	42.1	A	-	-	-
1	155140648	rs11264339	64,986	**AA**	**AB**		7997	8.8	7974	28.8	A	-	-	-
1	155205634	**N370S**	-	**-**	**-**		-	-	-	-	-	-	-	-
1	155218446	rs734074	-12,812	**AA**	**AB**		1132	0.7	7961	0.5	**A**	**A**	**A**	**N370S**
1	155224091	rs2075566	-18,457	**AA**	**AB**	67.7	7911	11.2	2115	34.8	**A**	**A**	**A**	**N370S**
1	155230131	rs1142287	-24,497	**AA**	**AB**		7804	41.4	7685	17.9	**A**	**A**	**A**	**N370S**
1	155273869	rs4971072	-68,235	**BB**	**AB**		5957	70.3	5353	55.6	**B**	**B**	**B**	**N370S**
1	155274960	rs12032720	-69,326	**AA**	**AB**		5848	1.8	7986	9.8	**A**	**A**	**A**	**N370S**
1	155282829	rs11264359	-77,195	**AA**	**AB**		7938	24.4	4796	1.6	**A**	**A**	**A**	**N370S**
1	155289545	rs11264361	-83,911	**AA**	**AB**		1602	31.8	1852	34.7	**A**	**A**	**A**	**N370S**
1	155318308	rs11264363	-112,674	**BB**	**AB**		1433	81.7	1862	62.3	**B**	**B**	**B**	**N370S**

1	155322142	rs12239114	-116,508	**AA**	**AB**		7944	0.9	3198	44.8	**A**	**A**	**A**	**N370S**

Abbreviations: MHiF, maternal haplotype in fetus; MFB N370S, maternal family-based N370S-linked haplotype; N370S cons, consensus N370S haplotype, FAI, fetal allele identity; DMAiF, diagnosed maternal allele in fetus; other abbreviations are the same as in Supplemental Table 6.

A. dbSNP ID or *GBA* mutation (red lettering)

B. For parental genotypes "AA" = homozygote dbSNP reference allele; "BB" = homozygote dbSNP non-reference allele; "AB" = heterozygote

C. Fetal load is 2x(mean paternal fetal fraction) as determined from SNP I and/or SNP II data (see Supplemental Methods)

D. B-allele frequency is the % frequency of (B-allele reads)/(total read depth (RD)) at the indicated nucleotide position

E. The maternal fetal haplotype (MHiF) was determined from SNP III data (as described in Supplemental Methods); the maternal N370S-linked haplotype (MFB N370S) was determined from family-based linkage analysis; the N370S consensus haplotype (N370S cons) was derived according to Figure 2. An "-" indicates that no haplotype data was available at the given position. Bold alleles were used for diagnosis of the maternal allele in the fetus ("DMAiF").

F. Fetal allele identity (FAI) was determined by comparing the "MHiF" haplotype to either the " MFB N370S" and/or "N370S cons" haplotypes

Supplemental Table 9. Identification of the maternal allele in the family 2 fetus (small panel)

Variant data							plasma DNA rep 2-1		plasma DNA rep 2-2		Haplotypes[E]			FAI[F]
Ch	Position (hg19)	dbSNP ID[A]	DFM (bps)	PGT[B]	MGT[B]	FL[C] (%)	RD (x)	BAF[D] (%)	RD (x)	BAF[D] (%)	MHiF	MFB N370S	N370S cons	DMAiF
1	155205634	N370S	-	-	-		-	-	-	-	-	-	-	-
1	155273869	rs4971072	-68,235	BB	AB	5.7	7915	86.2	2705	63.4	B	B	B	N370S
1	155282829	rs11264359	-77,195	AA	AB		7913	34.5	7929	30.1	A		A	N370S
1	155424065	rs11264375	-218,431	AA	AB		7945	27.3	6552	30.6	A		A	N370S

Abbreviations and footnotes are the same as in Supplemental Table 8.

Supplemental Table 10. Consensus Ashkenazi Jewish N370S founder haplotype

Ch	Position (hg19)	dbSNP ID	REF	ALT	consensus AJ N370S haplotype[A]
1	155,175,894	rs143461137	G	.	**A**
1	155,175,959	rs148003817	C	.	**A**
1	155,181,843	rs2974935	G	.	**A**
1	155,181,880	rs183507509	G	.	**A**
1	155,181,881	rs189399294	C	.	**A**
1	155,181,895	rs376771258	C	.	**A**
1	155,181,921	rs201017167	T	.	**A**
1	155,181,922	rs371111724	A	.	**A**
1	155,182,164	rs2075570	C	.	**A**
1	155,182,166	rs368470688	C	.	**A**
1	155,182,174	rs111443374	A	.	**A**
1	155,182,198	rs147593131	C	.	**A**
1	155,182,202	rs181961651	G	.	**A**
1	155,182,208	rs148055827	C	.	**A**
1	155,182,209	rs375049335	G	.	**A**
1	155,182,215	rs369259801	G	.	**A**
1	155,182,221	rs372693149	C	.	**A**
1	155,182,226	rs201939197	G	.	**A**
1	155,184,975	rs140081212	A	.	**A**
1	155,184,987	rs147747731	G	.	**A**
1	155,185,002	rs190092871	C	.	**A**
1	155,185,027	rs374532694	G	.	**A**
1	155,185,244	rs186678403	G	.	**A**
1	155,186,503	rs3125563	A	.	**A**
1	155,186,551	rs147791517	C	.	**A**
1	155,186,729	rs28445596	T	.	**A**
1	155,186,742	rs1057941	G	.	**A**
1	155,194,975	rs374166175	C	.	**A**
1	155,194,976	rs190676369	C	.	**A**
1	155,194,980	rs2049805	T	.	**A**
1	155,195,044	rs6677756	A	G	**B**
1	155,197,316	rs189462506	G	.	**A**
1	155,197,602	rs2990246	G	.	**A**
1	155,202,689	rs10796940	A	.	**A**
1	155,202,740	rs150410131	C	.	**A**
1	155,203,060	rs56310840	G	.	**A**
1	155,203,071	rs377284382	T	.	**A**
1	155,203,072	rs188784797	G	.	**A**
1	155,203,105	rs369352114	C	.	**A**
1	155,206,341	rs9628662	T	.	**A**
1	155,206,363	rs373282418	C	.	**A**
1	155,206,440	rs368332143	G	.	**A**
1	155,208,019	rs61748906	A	.	**A**

1	155,208,023	rs76727497	G	.	**A**
1	155,208,024	rs80205046	G	.	**A**
1	155,208,035	rs76717906	G	.	**A**
1	155,208,047	rs78659905	G	.	**A**
1	155,208,049	rs374591570	G	.	**A**
1	155,208,052	rs398123533	A	.	**A**
1	155,208,055	rs188760929	C	.	**A**
1	155,208,056	rs201615998	G	.	**A**
1	155,208,061	rs398123532	G	.	**A**
1	155,208,081	rs398123531	C	.	**A**
1	155,208,084	rs76500263	T	.	**A**
1	155,208,087	rs77933015	A	.	**A**
1	155,208,092	rs77916306	G	.	**A**
1	155,208,093	rs80222298	G	.	**A**
1	155,213,124	rs11264345	T	.	**A**
1	155,217,435	rs386635682	A	.	**A**
1	155,217,436	rs76718527	G	.	**A**
1	155,217,437	rs116352080	G	.	**A**
1	155,217,438	rs199735676	A	.	**A**
1	155,217,473	rs139111003	T	.	**A**
1	155,218,168	rs2075568	T	.	**A**
1	155,218,210	rs377201789	G	.	**A**
1	155,218,222	rs187706644	G	.	**A**
1	155,218,230	rs140120577	C	.	**A**
1	155,218,365	rs734073	T	.	**A**
1	155,218,370	rs139803480	C	.	**A**
1	155,218,446	rs734074	A	.	**A**
1	155,218,476	rs192678801	C	.	**A**
1	155,221,837	rs2072647	C	.	**A**
1	155,221,890	rs180894227	C	.	**A**
1	155,221,927	rs186280028	C	.	**A**
1	155,224,091	rs2075566	C	.	**A**
1	155,224,144	rs200790772	C	.	**A**
1	155,224,154	rs377616889	A	.	**A**
1	155,230,131	rs386580544	C	.	**A**
1	155,230,134	rs138266025	C	.	**A**
1	155,230,144	rs143921371	T	.	**A**
1	155,230,157	rs201400281	G	.	**A**
1	155,230,187	rs371041272	G	.	**A**
1	155,230,212	rs149415891	C	.	**A**
1	155,230,222	rs373833483	G	.	**A**
1	155,231,626	rs79051383	C	.	**A**
1	155,231,634	rs190472110	C	.	**A**
1	155,231,646	rs373599957	A	.	**A**
1	155,231,647	rs376455114	G	.	**A**
1	155,232,867	rs41264935	A	.	**A**
1	155,234,821	rs1076556	A	.	**A**
1	155,234,827	rs199775545	A	.	**A**
1	155,234,895	rs12044394	T	.	**A**

1	155,234,911	rs12043655	A	.	**A**
1	155,234,914	rs148772014	G	.	**A**
1	155,235,493	rs386515223	T	.	**A**
1	155,235,557	rs192046581	C	.	**A**
1	155,239,624	rs2361543	G	.	**A**
1	155,239,657	rs142820979	C	.	**A**
1	155,239,733	rs114256419	T	.	**A**
1	155,240,371	rs34467038	A	.	**A**
1	155,240,372	rs72461703	A	.	**A**
1	155,240,383	rs115310831	C	.	**A**
1	155,240,465	rs386596198	T	.	**A**
1	155,241,361	rs2236863	G	.	**A**
1	155,241,368	rs57336884	A	.	**A**
1	155,247,703	rs375207474	C	.	**A**
1	155,247,707	rs12749306	G	.	**A**
1	155,247,722	rs189401977	G	.	**A**
1	155,248,575	rs7549276	G	.	**A**
1	155,248,622	rs145609188	C	.	**A**
1	155,248,655	rs181989907	C	.	**A**
1	155,252,833	rs12049375	G	.	**A**
1	155,257,492	rs11264355	C	.	**A**
1	155,257,531	rs202118354	C	.	**A**
1	155,257,557	rs191318307	C	.	**A**
1	155,257,559	rs373694526	G	.	**A**
1	155,258,752	rs3814319	G	.	**A**
1	155,258,756	rs3814318	C	.	**A**
1	155,258,759	rs193130368	G	.	**A**
1	155,258,852	rs8177998	C	.	**A**
1	155,260,096	rs932972	G	.	**A**
1	155,260,348	rs374751259	G	.	**A**
1	155,260,350	rs1052177	A	.	**A**
1	155,260,360	rs368150636	G	.	**A**
1	155,260,382	rs61755431	C	.	**A**
1	155,260,383	rs1052176	G	.	**A**
1	155,260,402	rs140859641	G	.	**A**
1	155,260,415	rs370938255	C	.	**A**
1	155,260,434	rs370316462	C	.	**A**
1	155,260,443	rs190966689	C	.	**A**
1	155,260,448	rs375525211	C	.	**A**
1	155,279,550	rs144868148	C	.	**A**
1	155,282,829	rs11264359	A	.	**A**
1	155,282,884	rs34838448	T	.	**A**
1	155,289,545	rs11264361	T	.	**A**
1	155,289,607	rs201526312	G	.	**A**
1	155,289,616	rs201016550	G	.	**A**
1	155,289,623	rs371872906	C	.	**A**
1	155,289,626	rs146928362	C	.	**A**
1	155,318,308	rs11264363	G	C	**B**
1	155,318,313	rs138544252	T	.	**A**

1	155,318,353	rs368485885	T	.	**A**
1	155,322,142	rs12239114	G	.	**A**
1	155,322,147	rs148991117	T	.	**A**
1	155,322,211	rs187173286	C	.	**A**
1	155,345,043	rs5005770	A	.	**A**
1	155,345,056	rs374205358	T	.	**A**
1	155,345,077	rs73008959	G	.	**A**
1	155,425,259	rs200624751	C	.	**A**
1	155,425,261	rs201199578	A	.	**A**
1	155,425,263	rs202244553	T	.	**A**
1	155,429,490	rs10796946	A	.	**A**
1	155,429,496	rs148527468	C	.	**A**
1	155,429,526	rs377185391	C	.	**A**
1	155,429,538	rs368436478	T	.	**A**
1	155,429,543	rs34323458	A	.	**A**
1	155,429,548	rs113404715	G	.	**A**
1	155,429,725	rs10908466	A	.	**A**
1	155,429,806	rs369773358	T	.	**A**
1	155,431,171	rs35202668	C	.	**A**
1	155,431,172	rs200825345	G	.	**A**
1	155,431,173	rs201831219	T	.	**A**
1	155,431,193	rs115024233	T	.	**A**
1	155,433,942	rs12724079	T	C	**B**
1	155,433,971	rs140724364	G	.	**A**
1	155,434,017	rs114909331	A	.	**A**
1	155,466,784	rs12079134	C	.	**A**
1	155,468,732	rs10908469	A	.	**A**
1	155,473,356	rs1360554	C	T	**B**
1	155,478,861	rs75976369	A	.	**A**
1	155,478,897	rs6688636	T	C	**B**
1	155,501,014	rs35615695	T	C	**B**
1	155,501,096	rs80047375	A	.	**A**
1	155,508,882	rs6696888	G	A	**B**
1	155,508,901	rs188296881	T	.	**A**
1	155,508,922	rs72995173	C	.	**A**
1	155,509,107	rs10752611	C	.	**A**
1	155,522,572	rs11264381	T	.	**A**
1	155,522,603	rs149023559	C	.	**A**
1	155,527,013	rs10908470	T	.	**A**
1	155,558,665	rs3892792	G	.	**A**
1	155,558,738	rs372288696	A	.	**A**
1	155,558,761	rs141307650	C	.	**A**
1	155,559,170	rs11264386	G	.	**A**
1	155,559,174	rs372699570	T	.	**A**
1	155,559,185	rs676670	T	.	**A**
1	155,559,199	rs186071702	G	.	**A**
1	155,581,311	rs141265144	T	.	**A**
1	155,581,319	rs202139573	G	.	**A**
1	155,581,370	rs200351491	G	.	**A**

1	155,582,637	rs201559854	C	.	**A**
1	155,582,672	rs139011201	C	.	**A**
1	155,582,702	rs150075701	G	.	**A**
1	155,582,727	rs149262326	G	.	**A**
1	155,583,937	rs147713038	A	.	**A**
1	155,583,941	rs375845612	C	.	**A**
1	155,583,942	rs145476273	G	.	**A**
1	155,586,444	rs3851912	T	.	**A**
1	155,586,461	rs61258690	A	.	**A**
1	155,586,462	rs185906830	T	.	**A**
1	155,587,032	rs200023571	A	.	**A**
1	155,587,035	rs144981478	C	.	**A**
1	155,587,061	rs143206062	G	.	**A**
1	155,587,062	rs371733118	T	.	**A**
1	155,587,063	rs376673207	A	.	**A**
1	155,587,098	rs148274808	T	.	**A**
1	155,606,224	rs4971095	G	A	**B**
1	155,606,324	rs111692016	G	.	**A**
1	155,630,752	rs3738590	A	.	**A**
1	155,630,804	rs377670606	T	.	**A**
1	155,630,836	rs139714839	A	.	**A**
1	155,649,000	rs489970	A	.	**A**
1	155,652,081	rs475550	T	.	**A**
1	155,653,552	rs72508345	C	.	**A**
1	155,653,553	rs5777951	T	.	**A**
1	155,653,554	rs63024560	T	.	**A**
1	155,653,599	rs180689315	C	.	**A**
1	155,656,942	rs75256783	G	.	**A**
1	155,682,853	rs171166	A	.	**A**
1	155,682,868	rs186881907	G	.	**A**
1	155,682,870	rs56512938	C	.	**A**
1	155,682,898	rs139768145	T	.	**A**
1	155,682,917	rs375532832	G	.	**A**
1	155,687,232	rs621181	C	.	**A**
1	155,687,277	rs552778	G	.	**A**
1	155,719,161	rs112418368	T	.	**A**
1	155,719,466	rs386635696	G	.	**A**
1	155,721,474	rs596031	C	.	**A**
1	155,721,502	rs140892435	G	.	**A**
1	155,721,507	rs199974551	T	.	**A**
1	155,721,549	rs374668030	C	.	**A**
1	155,721,678	rs3863762	T	.	**A**
1	155,721,691	rs190512301	G	.	**A**
1	155,722,334	rs623440	C	.	**A**
1	155,722,345	rs374300283	C	.	**A**
1	155,722,364	rs34391545	T	.	**A**
1	155,722,395	rs377411800	T	.	**A**
1	155,722,403	rs34838598	T	.	**A**
1	155,735,675	rs676814	A	.	**A**

1	155,735,688	rs201627893	A	.	**A**
1	155,735,698	rs267598068	G	.	**A**
1	155,735,721	rs677219	A	.	**A**
1	155,735,723	rs677220	T	.	**A**
1	155,735,726	rs200987293	T	.	**A**
1	155,735,732	rs371491793	T	.	**A**
1	155,735,752	rs142382216	A	.	**A**
1	155,735,790	rs150057388	G	.	**A**
1	155,737,054	rs146396042	C	.	**A**
1	155,737,055	rs487451	A	.	**A**
1	155,737,101	rs488237	T	.	**A**
1	155,737,104	rs488240	A	.	**A**
1	155,737,118	rs202234781	A	.	**A**
1	155,737,147	rs488349	C	.	**A**
1	155,743,310	rs708611	A	C	**B**
1	155,744,487	rs143645681	G	.	**A**
1	155,744,509	rs112412750	C	.	**A**
1	155,753,676	rs822019	A	G	**B**
1	155,753,727	rs371575442	G	.	**A**
1	155,753,742	rs373602803	T	.	**A**
1	155,764,808	rs3738591	C	.	**A**
1	155,764,854	rs142487782	T	.	**A**
1	155,764,893	rs376934806	G	.	**A**
1	155,783,762	rs200898321	T	.	**A**
1	155,783,763	rs822012	C	T	**B**
1	155,783,830	rs115743054	A	.	**A**
1	155,783,882	rs192732599	G	.	**A**
1	155,793,689	rs822477	C	A	**B**
1	155,793,752	rs377224351	C	.	**A**
1	155,793,757	rs149516593	A	.	**A**
1	155,810,293	rs822497	T	C	**B**
1	155,810,323	rs369365982	G	.	**A**
1	155,822,971	rs822490	C	T	**B**
1	155,822,974	rs201919208	C	.	**A**
1	155,823,002	rs822491	T	C	**B**
1	155,823,029	rs372837679	T	.	**A**
1	155,823,033	rs41264979	C	.	**A**
1	155,823,050	rs369296856	G	.	**A**
1	155,823,078	rs139134396	T	.	**A**
1	155,829,510	rs375004581	T	.	**A**
1	155,829,511	rs3820594	C	.	**A**
1	155,829,535	rs371110583	A	.	**A**
1	155,829,551	rs373914518	A	.	**A**
1	155,829,573	rs144636810	C	.	**A**
1	155,829,584	rs377646230	T	.	**A**
1	155,840,706	rs822519	C	T	**B**
1	155,840,764	rs191397830	C	.	**A**
1	155,840,783	rs79096273	T	.	**A**
1	155,840,788	rs182565567	T	.	**A**

1	155,843,254	rs192881572	T	.	**A**
1	155,843,256	rs7528441	T	.	**A**
1	155,843,318	rs184328077	T	.	**A**
1	155,846,528	rs822505	C	T	**B**
1	155,846,601	rs143502811	G	.	**A**
1	155,850,558	rs822508	T	C	**B**
1	155,850,585	rs192002378	T	.	**A**
1	155,850,588	rs183430264	C	.	**A**
1	155,850,811	rs822509	G	A	**B**
1	155,850,819	rs822510	C	.	**A**
1	155,850,822	rs187974322	A	.	**A**
1	155,850,889	rs192773635	C	.	**A**
1	155,868,625	rs2282301	G	.	**A**

Abbreviations: Ch, chromosome; REF, reference nucleotide (dbSNP Build 141); ALT, alternate (non-reference) nucleotide (dbSNP Build 141)

A. For the consensus AJ N370S haplotype: "A" = dbSNP reference nucleotide; "B" = dbSNP non-reference nucleotide

B. The region shaded in gray indicates *GBA* intragenic loci

Supplemental Table 11. Parental family-based haplotype information (from large sequencing panel)

Family	Paternal familial haplotype data				Maternal familial haplotype data			
	Paternal genotype	Paternal family member/s used for linkage	Genotype of paternal family member	No. of SNPs in linked haplotype	Maternal genotype	Maternal family member/s used for linkage	Genotype of maternal family member	No. of SNPs in linked haplotype
1	N370S/WT	father	N370S/WT	271	N370S/WT	sister	N370S/WT	231
2	WT/WT	N/A	N/A	N/A	N370S/WT	mother	N370S/WT	113
3	WT/WT	N/A	N/A	N/A	N370S/V394L	mother	V394L/WT	201
						father	N370S/WT	325
4	R496H/WT	mother	R496H/WT	309	N370S/WT	mother	N370S/WT	270
		father	WT/WT	336		father	WT/WT	314
5	N370S/del55	mother	del55/WT	302	WT/WT	N/A	N/A	N/A
		father	N370S/WT	191				
6	N370S/84GG	mother	N370S/WT	269	WT/WT	N/A	N/A	N/A
		brother	N370S/WT	327				
7	WT/WT	N/A	N/A	N/A	N370S/L444P	mother	L444P/WT	225
						father	N370S/WT	291

8	WT/WT	N/A	N/A	N/A	N370S/84GG	mother	N370S/WT	321
						father	84GG/WT	186

Abbreviations: WT, wild type; N/A, not applicable

Supplemental Table 12. Identification of the paternal allele in the family 1 fetus (large panel)

Variant data							plasma DNA rep 1-1		plasma DNA rep 1-2		Haplotypes[E]			FAI[F]
Ch	Position (hg19)	dbSNP ID[A]	DFM (bps)	PGT[B]	MGT[B]	FL[C] (%)	RD (x)	BAF[D]	RD (x)	BAF[D]	PHiF	PFB N370S	N370S cons	DPAiF
1	153,227,177	rs873234	1,978,457	**AB**	**BB**		510	**95.5%**	442	98.6%	**A**	**B**	-	**WT**
1	153,408,831	rs2026604	1,796,803	**AB**	**AA**		2836	**2.0%**	603	1.3%	B	-	-	-
1	153,433,936	rs12049559	1,771,698	**AB**	**AA**		2978	**46.2%**	7918	**24.0%**	**B**	**A**	-	**WT**
1	153,636,860	rs4351684	1,568,774	**AB**	**AA**		7988	**17.2%**	7867	1.5%	B	-	-	**-**
1	153,779,412	rs1127091	1,426,222	**AB**	**BB**		2225	99.8%	3359	**96.3%**	A	-	-	**-**
1	153,780,298	rs9426938	1,425,336	**AB**	**BB**		347	**95.4%**	755	98.1%	A	-	-	-
1	153,886,762	rs12032914	1,318,872	**AB**	**AA**	13.6	1308	**22.7%**	867	1.2%	B	-	-	-
1	153,901,149	rs3748849	1,304,485	**AB**	**AA**		114	0.9%	582	**24.6%**	B	-	-	-
1	153,927,052	rs10908557	1,278,582	**AB**	**AA**		2079	**9.0%**	2677	0.3%	B	-	-	-
1	154,150,911	rs12063830	1,054,723	**AB**	**AA**		752	**24.5%**	5448	**44.3%**	B	-	-	-
1	154,302,826	rs6691345	902,808	**AB**	**AA**		5003	1.3%	7385	**2.9%**	**B**	**A**	-	**WT**
1	154,791,676	rs386523173	413,958	**AB**	**AA**		7928	0.2%	7905	**7.1%**	**B**	**A**	-	**WT**
1	154,792,269	rs2798603	413,365	**AB**	**AA**		1527	**1.7%**	2536	1.2%	**B**	**A**	-	**WT**

1	155,019,710	rs11264300	185,924	**AB**	**BB**	385	99.7%	1001	**97.6%**	A	-	-	-
1	155,205,634	**N370S**	-	**-**	**-**	-	-	-	-	-	-	-	-
1	156,230,936	rs759329	-1,025,302	**AB**	**AA**	7941	0.9%	4733	**12.4%**	B	-	-	-
1	156,244,992	rs2270291	-1,039,358	**AB**	**AA**	3598	**3.6%**	1703	0.3%	**B**	**A**	-	**WT**
1	156,251,742	rs3806407	-1,046,108	**AB**	**AA**	1848	0.2%	2568	**6.3%**	**B**	**A**	-	**WT**
1	156,299,916	rs12048077	-1,094,282	**AB**	**AA**	665	1.1%	712	**2.4%**	**B**	**A**	-	**WT**
1	156,309,372	rs2296375	-1,103,738	**AB**	**AA**	312	0.3%	1195	**24.5%**	**B**	**A**	-	**WT**
1	156,387,167	rs16837375	-1,181,533	**AB**	**AA**	7940	**3.4%**	7943	0.3%	**B**	**A**	-	**WT**
1	156,799,912	rs12756019	-1,594,278	**AB**	**BB**	862	99.5%	532	**98.9%**	**A**	**B**	-	**WT**
1	157,088,386	rs4661088	-1,882,752	**AB**	**AA**	1030	**6.9%**	1562	**3.2%**	**B**	**A**	-	**WT**
1	157,090,136	rs12022068	-1,884,502	**AB**	**AA**	7895	**3.0%**	3776	0.4%	**B**	**A**	-	**WT**
1	157,090,760	rs11264618	-1,885,126	**AB**	**AA**	6462	0.6%	2507	**2.9%**	**B**	**A**	-	**WT**
1	157,134,406	rs1176548	-1,928,772	**AB**	**BB**	12322	1.3%	1218	**66.7%**	**A**	**B**	-	**WT**
1	157,195,934	rs17415227	-1,990,300	**AB**	**AA**	3857	**6.1%**	372	**31.7%**	B	-	-	-

Abbreviations and footnotes are the same as in Supplemental Table 6.

Supplemental Table 13. Identification of the maternal allele in the family 1 fetus (large panel)

Variant data							plasma DNA rep 1-1		plasma DNA rep 1-2		Haplotypes[E]			FAI[F]
Ch	Position (hg19)	dbSNP ID[A]	DFM (bps)	PGT[B]	MGT[B]	FL[C] (%)	RD (x)	BAF[D]	RD (x)	BAF[D]	MHiF	MFB N370S	N370S cons	DMAiF
1	153,334,525	rs1560833	1,871,109	**AA**	**AB**		894	37.2%	571	17.5%	A	-	-	-
1	153,637,404	rs6427671	1,568,230	**BB**	**AB**		174	61.5%	381	99.5%	B	-	-	-
1	154,239,283	rs1212352	966,351	**AA**	**AB**		14973	1.5%	6539	13.1%	**A**	**A**	-	**N370S**
1	154,259,275	rs1760802	946,359	**AA**	**AB**		1499	3.3%	846	8.2%	**A**	**A**	-	**N370S**
1	154,591,882	rs56020456	613,752	**AA**	**AB**		957	1.5%	1979	22.7%	A	-	-	-
1	154,688,197	rs11264248	517,437	**AA**	**AB**		11237	19.9%	7794	14.8%	A	-	-	-
1	154,910,012	rs877343	295,622	**BB**	**AB**	13.6	137	97.1%	169	74.0%	B	-	-	-
1	155,047,719	rs11264307	157,915	**AA**	**AB**		1125	1.0%	605	0.5%	A	-	-	-
1	155,079,477	rs7367207	126,157	**AA**	**AB**		845	30.1%	179	35.2%	**A**	-	**A**[G]	**N370S**
1	155,135,335	rs4460629	70,299	**AA**	**AB**		212	0.9%	279	6.1%	**A**	-	**A**[G]	**N370S**
1	155,140,648	rs11264339	64,986	**AA**	**AB**		7943	9.3%	7932	19.6%	**A**	-	**A**[G]	**N370S**
1	155,205,634	**N370S**	-	**-**	**-**		-	-	-	-	-	-	-	-
1	155,213,124	rs11264345	-7,490	**AA**	**AB**		7706	36.5%	5395	2.3%	**A**	**A**	**A**	**N370S**

1	155,224,091	rs2075566	-18,457	**AA**	**AB**	1825	10.0%	1832	20.4%	**A**	**A**	**A**	**N370S**
1	155,230,131	rs386580544	-24,497	**AA**	**AB**	7781	40.2%	7691	16.7%	**A**	**A**	**A**	**N370S**
1	155,235,493	rs386515223	-29,859	**AA**	**AB**	1462	29.8%	758	3.8%	**A**	**A**	**A**	**N370S**
1	155,239,624	rs2361543	-33,990	**AA**	**AB**	1110	23.5%	525	0.2%	**A**	**A**	**A**	**N370S**
1	155,282,829	rs11264359	-77,195	**AA**	**AB**	7969	20.9%	7951	2.3%	**A**	**A**	**A**	**N370S**
1	155,322,142	rs12239114	-116,508	**AA**	**AB**	950	0.8%	167	40.1%	**A**	**A**	**A**	**N370S**
1	155,429,725	rs10908466	-224,091	**AA**	**AB**	1531	1.8%	14738	4.2%	**A**	**A**	**A**	**N370S**
1	155,630,752	rs3738590	-425,118	**AA**	**AB**	7941	1.2%	7866	28.9%	**A**	**A**	**A**	**N370S**
1	155,822,971	rs822490	-617,337	**BB**	**AB**	690	97.7%	224	72.3%	**B**	**B**	**B**	**N370S**
1	156,055,099	rs12076700	-849,465	**AA**	**AB**	1602	19.1%	1549	27.5%	**A**	**A**	-	**N370S**
1	156,055,185	rs12063564	-849,551	**AA**	**AB**	8061	2.6%	2224	27.2%	**A**	**A**	-	**N370S**
1	156,650,909	rs11264516	-1,445,275	**AA**	**AB**	130	0.0%	135	0.7%	A	-	-	-
1	156,678,291	rs67794440	-1,472,657	**AA**	**AB**	256	1.2%	2816	1.4%	A	-	-	-

Abbreviations and footnotes are the same as in Supplemental Table 8 with the following additional footnote:

G. Near consensus N370S haplotype as determined according to Figure 4.

Supplemental Table 14. Identification of the maternal allele in the family 2 fetus (large panel)

Variant data							plasma DNA rep 2-1		plasma DNA rep 2-2		Haplotypes[E]			FAI[F]
Ch	Position (hg19)	dbSNP ID[A]	DFM (bps)	PGT[B]	MGT[B]	FL[C] (%)	RD (x)	BAF[D]	RD (x)	BAF[D]	MHiF	MFB N370S	N370S cons	DMAiF
1	153,283,495	rs2987760	1,922,139	**BB**	**AB**		309	99.7%	163	98.8%	B	-	-	-
1	153,283,978	rs821420	1,921,656	**AA**	**AB**		2890	2.7%	930	1.8%	A	-	-	-
1	153,334,525	rs1560833	1,871,109	**AA**	**AB**		7311	17.9%	504	6.3%	**A**	**A**	-	**N370S**
1	153,388,881	rs3014812	1,816,753	**AA**	**AB**		7542	13.1%	2609	2.1%	A	-	-	-
1	153,475,660	rs16835382	1,729,974	**BB**	**AB**		3752	65.2%	1492	84.6%	B	-	-	-
1	153,591,652	rs9726753	1,613,982	**AA**	**AB**		900	4.2%	389	39.8%	**A**	**A**	-	**N370S**
1	154,192,143	rs9803862	1,013,491	**AA**	**AB**	9.2	186	27.4%	1086	11.2%	A	-	-	-
1	154,206,212	rs10797061	999,422	**AA**	**AB**		644	1.9%	873	8.9%	A	-	-	-
1	154,219,485	rs1194580	986,149	**AA**	**AB**		169	0.6%	459	0.4%	A	-	-	-
1	154,219,718	rs6427509	985,916	**AA**	**AB**		4968	17.7%	6617	0.3%	A	-	-	-
1	154,308,680	rs9787014	896,954	**AA**	**AB**		7906	24.3%	677	34.4%	A	-	-	-
1	154,310,048	rs1760795	895,586	**AA**	**AB**		193	0.0%	668	32.0%	A	-	-	-
1	154,411,419	rs4845622	794,215	**AA**	**AB**		2041	1.6%	192	15.6%	A	-	-	-

1	154,554,357	rs11264222	651,277	**AA**	**AB**	4701	30.8%	1445	36.4%	**A**	**A**	-	**N370S**
1	154,573,967	rs386561757	631,667	**BB**	**AB**	7746	98.9%	167	100.0%	**B**	**B**	-	**N370S**
1	154,677,716	rs56320421	527,918	**AA**	**AB**	7966	44.9%	7070	0.7%	A	-	-	-
1	154,678,537	rs61811430	527,097	**AA**	**AB**	7971	29.1%	7869	24.2%	A	-	-	-
1	154,688,197	rs11264248	517,437	**BB**	**AB**	7856	78.8%	7827	74.0%	B	-	-	-
1	154,790,480	rs35612271	415,154	**AA**	**AB**	7898	27.6%	2431	2.6%	A	-	-	-
1	155,205,634	**N370S**	-	**-**	**-**	-	-	-	-	-	-	-	-
1	155,282,829	rs11264359	-77,195	**AA**	**AB**	7964	32.8%	7960	25.9%	**A**	-	**A**	**N370S**
1	155,433,942	rs12724079	-228,308	**BB**	**AB**	539	63.3%	103	96.1%	**B**	-	**B**	**N370S**
1	155,466,784	rs12079134	-261,150	**AA**	**AB**	2129	27.1%	420	0.0%	**A**	-	**A**	**N370S**
1	155,846,528	rs822505	-640,894	**BB**	**AB**	1125	99.1%	717	95.7%	**B**	-	**B**	**N370S**
1	155,880,159	rs867549	-674,525	**AA**	**AB**	7863	31.7%	5350	1.4%	**A**	-	**A**^{G}	**N370S**
1	156,287,360	rs2249790	-1,081,726	**AA**	**AB**	648	36.7%	7901	0.3%	**A**	**A**	**A**^{G}	**N370S**
1	157,104,470	rs6704373	-1,898,836	**AA**	**AB**	7847	0.6%	169	22.5%	A	-	-	-
1	157,113,029	rs7544727	-1,907,395	**AA**	**AB**	2306	0.7%	275	0.0%	A	-	-	-

Abbreviations and footnotes are the same as in Supplemental Table 13.

Supplemental Table 15. Identification of the maternal allele in the family 3 fetus (large panel)

Variant data							plasma DNA rep 3-1		plasma DNA rep 3-2		Haplotypes[E]				FAI[F]
Ch	Position (hg19)	dbSNP ID[A]	DFM (bps)	PGT[B]	MGT[B]	FL[C] (%)	RD (x)	BAF[D]	RD (x)	BAF[D]	MHiF	MFB N370S	MFB V394L	N370S cons	DMAiF
1	153,221,315	rs12131158	1,984,319	**AA**	**AB**		4234	1.5%	7937	1.5%	A	-	-	-	-
1	153,327,732	rs2916198	1,877,902	**AA**	**AB**		590	32.9%	1367	28.3%	**A**	**A**	-	-	**N370S**
1	154,239,283	rs1212352	966,351	**AA**	**AB**		7897	24.9%	7908	9.3%	A	-	-	-	-
1	154,325,149	rs2483712	880,485	**BB**	**AB**		7899	88.8%	4805	80.6%	**B**	**B**	-	-	**N370S**
1	154,426,947	rs8192283	778,687	**AA**	**AB**		3541	36.8%	3500	41.9%	**A**	**A**	-	-	**N370S**
1	154,451,420	rs11265622	754,214	**BB**	**AB**		2848	68.2%	6497	75.8%	**B**	**B**	-	-	**N370S**
1	154,514,331	rs4845647	691,303	**BB**	**AB**	3.4	1823	65.1%	2381	78.7%	**B**	**B**	-	-	**N370S**
1	154,514,338	rs4845648	691,296	**AA**	**AB**		1769	33.4%	2291	18.3%	**A**	**A**	-	-	**N370S**
1	154,682,082	rs10908427	523,552	**AA**	**AB**		1009	3.6%	526	6.7%	**A**	-	**B**	-	**N370S**
1	154,760,595	rs1777910	445,039	**BB**	**AB**		3106	67.3%	1292	71.4%	**B**	**B**	-	-	**N370S**
1	154,779,122	rs906274	426,512	**AA**	**AB**		7968	0.7%	3157	1.0%	**A**	**A**	-	-	**N370S**
1	154,784,466	rs7513480	421,168	**AA**	**AB**		7812	31.9%	2676	27.5%	**A**	**A**	**B**	-	**N370S**
1	155,205,634	**N370S**	-	**-**	**-**		-	-	-	-	-	-	-	-	-

1	156,127,368	rs12128066	-921,734	**AA**	**AB**	7559	11.7%	948	41.1%	**A**	**A**	-	-	**N370S**
1	156,382,470	rs12401436	-1,176,836	**AA**	**AB**	1435	6.1%	2465	42.2%	**A**	**A**	-	-	**N370S**
1	156,387,167	rs16837375	-1,181,533	**AA**	**AB**	7966	35.9%	7974	28.4%	**A**	**A**	-	-	**N370S**
1	156,701,372	rs73000798	-1,495,738	**AA**	**AB**	5135	28.5%	7915	8.7%	**A**	**A**	-	-	**N370S**

Abbreviations and footnotes are the same as in Supplemental Table 13 with the following footnote and abbreviation modifications:

E. The maternal fetal haplotype (MHiF) was determined from SNP III data (as described in Supplemental Methods); the maternal N370S-linked (MFB N370S) and maternal V394L-linked (MFB V394L) haplotypes were determined by family-based linkage analysis; the N370S consensus haplotype (N370S cons) was derived according to Figure 2. An "-" indicates that no haplotype data was available at the given position. Bold alleles were used for diagnosis of the maternal allele in the fetus ("DMAiF").

F. Fetal allele identity (FAI) was determined by comparing the "MHiF" haplotype to the "MFB N370S", "MFB V394L", and/or "N370S cons" haplotypes

Supplemental Table 16. Identification of the paternal allele in the family 4 fetus (large panel)

Variant data							plasma DNA rep 4-1		plasma DNA rep 4-2		Haplotypes[E]			FAI[F]
Ch	Position (hg19)	dbSNP ID[A]	DFM (bps)	PGT[B]	MGT[B]	FL[C] (%)	RD (x)	BAF[D]	RD (x)	BAF[D]	PHiF	PFB R496H	PFB WT	DPAiF
1	154,464,572	rs4478801	740,221	**AB**	**AA**		7963	**3.2%**	7954	0.3%	B	-	-	-
1	154,492,432	rs4518898	712,361	**AB**	**BB**		1599	**98.9%**	561	99.8%	**A**	**B**	-	**WT**
1	154,526,414	rs7539745	678,379	**AB**	**BB**		7626	100.0%	7725	**98.7%**	**A**	**B**	-	**WT**
1	154,774,966	rs6690891	429,827	**AB**	**AA**		7696	1.1%	7552	**2.1%**	**B**	**A**	-	**WT**
1	155,186,742	rs1057941	18,051	**AB**	**AA**		3226	0.8%	2770	**2.1%**	**B**	**A**	**B**	**WT**
1	155,197,602	rs2990246	7,191	**AB**	**AA**		3625	**1.7%**	2985	0.2%	**B**	**A**	-	**WT**
1	155,204,793	**R496H**	-	**-**	**-**	5.6	-	-	-	-	-	-	-	-
1	155,581,311	rs141265144	-376,518	**AB**	**AA**		7889	0.5%	7850	**5.9%**	B	-	-	-
1	156,230,546	rs12142808	-1,025,753	**AB**	**AA**		2931	1.1%	3520	**1.7%**	**B**	-	**B**	**WT**
1	156,314,147	rs12022657	-1,109,354	**AB**	**AA**		721	0.8%	939	**2.6%**	**B**	-	**B**	**WT**
1	156,618,768	rs4661209	-1,413,975	**AB**	**BB**		675	**95.1%**	1008	97.9%	**A**	**B**	-	**WT**
1	156,623,625	rs10908513	-1,418,832	**AB**	**BB**		4848	**98.0%**	7440	99.1%	A	-	-	-
1	156,671,469	rs12406221	-1,466,676	**AB**	**BB**		1009	**94.3%**	1970	99.4%	**A**	-	**A**	**WT**

1	157,061,220	rs6677868	-1,856,427	**AB**	**BB**	5358	**62.5%**	7764	**93.2%**	**A**	**B**	**A**	**WT**
1	157,135,165	rs1176550	-1,930,372	**AB**	**BB**	6826	98.9%	7138	**95.4%**	**A**	**B**	-	**WT**
1	157,137,112	rs1176556	-1,932,319	**AB**	**BB**	696	**96.1%**	624	99.8%	**A**	**B**	-	**WT**

Abbreviations and footnotes are the same as in Supplemental Table 6 with the following footnote and abbreviation modifications:

E. The paternal fetal haplotype (PHiF) was determined from SNP II data (as described in Supplemental Methods); the paternal R496H-linked (PFB R496H) and wild type-linked (PFB WT) haplotypes were determined from family-based linkage analysis. An "-" indicates that no haplotype data was available at the given position. Bold alleles were used for diagnosis of the paternal allele in the fetus ("DPAiF").

F. Fetal allele identity (FAI) was determined by comparing the "PHiF" haplotype to the "PFB R496H" and/or “PFB WT” haplotypes.

Supplemental Table 17. Identification of the maternal allele in the family 4 fetus (large panel)

Variant data							plasma DNA rep 4-1		plasma DNA rep 4-2		Haplotypes[E]				FAI[F]
Ch	Position (hg19)	dbSNP ID[A]	DFM (bps)	PGT[B]	MGT[B]	FL[C] (%)	RD (x)	BAF[D]	RD (x)	BAF[D]	MHiF	MFB N370S	MFB WT	N370S cons	DMAiF
1	153,218,349	rs11583609	1,987,285	**AA**	**AB**		111	4.5%	579	27.8%	**A**	-	**A**	-	**WT**
1	153,221,635	rs12760683	1,983,999	**AA**	**AB**		374	5.3%	348	26.1%	**A**	-	**A**	-	**WT**
1	153,266,256	rs11585177	1,939,378	**AA**	**AB**		4743	39.6%	7773	42.0%	**A**	-	**A**	-	**WT**
1	153,293,934	rs2916230	1,911,700	**AA**	**AB**		539	19.1%	569	32.0%	**A**	-	**A**	-	**WT**
1	153,294,653	rs2916229	1,910,981	**AA**	**AB**		891	19.3%	690	35.5%	**A**	-	**A**	-	**WT**
1	153,309,981	rs12139503	1,895,653	**AA**	**AB**		7918	19.2%	6346	28.8%	**A**	-	**A**	-	**WT**
1	153,753,725	rs10908474	1,451,909	**AA**	**AB**	5.6	1983	40.3%	817	41.1%	A	-	-	-	-
1	153,833,591	rs7552274	1,372,043	**AA**	**AB**		1744	35.8%	3921	33.1%	A	-	-	-	-
1	153,851,106	rs12041920	1,354,528	**AA**	**AB**		610	35.9%	1043	18.5%	A	-	-	-	-
1	153,901,149	rs3748849	1,304,485	**AA**	**AB**		987	23.3%	1532	7.5%	A	-	-	-	-
1	153,919,756	rs946682	1,285,878	**AA**	**AB**		4790	27.9%	4746	30.7%	A	-	-	-	-
1	153,926,078	rs11264680	1,279,556	**AA**	**AB**		1533	14.5%	708	38.0%	A	-	-	-	-
1	153,992,734	rs12048137	1,212,900	**AA**	**AB**		7933	23.3%	4701	16.6%	A	-	-	-	-

1	154,692,883	rs7554577	512,751	**AA**	**AB**	1228	19.5%	1696	43.6%	A	-	-	-	-
1	154,796,520	rs11264268	409,114	**AA**	**AB**	3522	41.9%	6169	24.9%	A	-	-	-	-
1	154,822,517	rs906597	383,117	**AA**	**AB**	1924	21.7%	5691	9.9%	**A**	-	**A**	-	**WT**
1	155,106,054	rs4390169	99,580	**AA**	**AB**	763	31.8%	945	15.2%	**A**	**B**	-	**B^{G}**	**WT**
1	155,106,227	rs4745	99,407	**AA**	**AB**	7376	37.3%	6730	19.6%	**A**	**B**	-	**B^{G}**	**WT**
1	155,205,634	**N370S**	-	**-**	**-**	-	-	-	-	-	-	-	-	-
1	155,870,416	rs1749409	-664,782	**BB**	**AB**	593	62.6%	245	56.3%	**B**	-	-	**A^{G}**	**WT**
1	155,917,880	rs1010033	-712,246	**BB**	**AB**	373	93.8%	336	66.4%	**B**	-	-	**A^{G}**	**WT**
1	156,089,920	rs610918	-884,286	**AA**	**AB**	1400	7.1%	5112	8.7%	**A**	-	-	**B^{G}**	**WT**
1	156,093,195	rs693671	-887,561	**AA**	**AB**	7743	18.5%	6812	25.9%	**A**	-	-	**B^{G}**	**WT**
1	156,099,830	rs666869	-894,196	**AA**	**AB**	368	5.4%	1429	2.8%	**A**	-	-	**B^{G}**	**WT**
1	156,106,863	rs553016	-901,229	**AA**	**AB**	364	37.9%	170	18.8%	**A**	-	-	**B^{G}**	**WT**
1	156,117,584	rs72708271	-911,950	**AA**	**AB**	611	12.1%	1677	37.2%	**A**	-	-	**B^{G}**	**WT**
1	156,241,701	rs2246476	-1,036,067	**AA**	**AB**	7916	26.6%	3974	31.9%	**A**		**A**	**B^{G}**	**WT**
1	156,246,473	rs7542045	-1,040,839	**BB**	**AB**	3250	67.4%	3173	74.5%	**B**	**A**	**B**	**A^{G}**	**WT**
1	156,508,477	rs1177696	-1,302,843	**BB**	**AB**	7873	54.5%	7895	86.4%	**B**	**A**	-	-	**WT**
1	156,518,379	rs744224	-1,312,745	**BB**	**AB**	1548	62.1%	1235	59.8%	**B**	**A**	-	-	**WT**
1	156,519,799	rs1778820	-1,314,165	**BB**	**AB**	144	65.3%	312	94.9%	**B**	**A**	-	-	**WT**

1	156,534,539	rs4661185	-1,328,905	**AA**	**AB**	6599	14.0%	7342	18.5%	**A**	**B**	-	-	**WT**
1	156,626,508	rs34478535	-1,420,874	**AA**	**AB**	1427	34.0%	2207	31.2%	A	-	-	-	-
1	156,645,948	rs77658164	-1,440,314	**AA**	**AB**	226	32.3%	408	17.4%	A	-	-	-	-
1	156,660,311	rs4384231	-1,454,677	**AA**	**AB**	956	21.8%	114	12.3%	**A**	-	**A**	-	**WT**
1	157,194,156	rs12126856	-1,988,522	**AA**	**AB**	4868	39.7%	6041	31.7%	**A**	-	**A**	-	**WT**
1	157,194,578	rs11264636	-1,988,944	**AA**	**AB**	994	20.5%	280	27.9%	**A**	-	**A**	-	**WT**
1	157,196,031	rs10908543	-1,990,397	**AA**	**AB**	343	8.5%	242	5.8%	**A**	-	**A**	-	**WT**

Abbreviations and footnotes are the same as in Supplemental Table 13 with the following footnote and abbreviation modifications:

E. The maternal fetal haplotype (MHiF) was determined from SNP III data (as described in Supplemental Methods); the maternal N370S-linked (MFB N370S) and maternal wild type-linked (MFB WT) haplotypes were determined by family-based linkage analysis; the N370S consensus haplotype (N370S cons) was derived according to Figure 2. An "-" indicates that no haplotype data was available at the given position. Bold alleles were used for diagnosis of the maternal allele in the fetus ("DMAiF").

F. Fetal allele identity (FAI) was determined by comparing the "MHiF" haplotype to the "MFB N370S", "MFB WT", and/or "N370S cons" haplotypes

Supplemental Table 18. Identification of the paternal allele in the family 5 fetus (large panel)

Variant data							plasma DNA rep 5-1		plasma DNA rep 5-2		Haplotypes[E]				FAI[F]
Ch	Position (hg19)	dbSNP ID[A]	DFM (bps)	PGT[B]	MGT[B]	FL[C] (%)	RD (x)	BAF[D]	RD (x)	BAF[D]	PHiF	PFB N370S	PFB del55	N370S cons	DPAiF
1	153,609,039	rs4845557	1,596,595	**AB**	**BB**		656	**98.3%**	986	**96.6%**	A	-	-	-	-
1	153,612,053	rs2274740	1,593,581	**AB**	**BB**		714	99.6%	558	**98.6%**	A	-	-	-	-
1	153,622,220	rs913861	1,583,414	**AB**	**BB**		606	**93.2%**	684	**98.0%**	A	-	-	-	-
1	153,636,860	rs4351684	1,568,774	**AB**	**BB**		7980	**96.0%**	7878	**98.6%**	A	-	-	-	-
1	153,869,946	rs12401565	1,335,688	**AB**	**AA**		3237	0.3%	1118	**5.2%**	**B**	-	**A**	-	**N370S**
1	153,908,309	rs11586075	1,297,325	**AB**	**AA**		2812	1.2%	4687	**5.9%**	**B**	-	**A**	-	**N370S**
1	153,919,756	rs946682	1,285,878	**AB**	**AA**	5.8	7978	0.9%	7974	**1.4%**	**B**	-	**A**	-	**N370S**
1	153,927,052	rs10908557	1,278,582	**AB**	**AA**		7887	**2.1%**	6059	1.1%	**B**	-	**A**	-	**N370S**
1	153,932,212	rs6724	1,273,422	**AB**	**AA**		7911	**1.4%**	5776	0.7%	**B**	-	**A**	-	**N370S**
1	153,986,637	rs11264865	1,218,997	**AB**	**AA**		7370	**8.3%**	1819	0.4%	**B**	-	**A**	-	**N370S**
1	154,017,501	rs2841102	1,188,133	**AB**	**AA**		7867	**2.1%**	7315	**4.2%**	**B**	-	**A**	-	**N370S**
1	154,669,294	rs386559666	536,340	**AB**	**BB**		2946	99.4%	5467	**98.5%**	**A**	**A**	**B**	-	**N370S**
1	154,797,062	rs6681725	408,572	**AB**	**AA**		694	1.2%	890	**2.0%**	**B**	-	**A**	-	**N370S**

1	155,205,634	**N370S**	-	**-**	**-**	-	-	-	-	-	-	-	-	-
1	155,983,648	rs17381047	-778,014	**AB**	**BB**	6262	99.2%	4973	**97.5%**	**A**	**A**	**B**	**A^{G}**	**N370S**
1	156,011,444	rs2297792	-805,810	**AB**	**BB**	7978	**97.9%**	7979	99.5%	**A**	**A**	**B**	**A^{G}**	**N370S**
1	156,024,373	rs3754293	-818,739	**AB**	**BB**	3377	99.7%	1752	**97.8%**	**A**	**A**	**B**	**A^{G}**	**N370S**
1	156,230,936	rs759329	-1,025,302	**AB**	**BB**	7977	99.5%	7985	**97.8%**	**A**	**A**	**B**	**A^{G}**	**N370S**
1	156,394,000	rs7548323	-1,188,366	**AB**	**AA**	784	**1.3%**	957	0.3%	**B**	-	**A**	-	**N370S**
1	156,446,450	rs12131289	-1,240,816	**AB**	**BB**	549	**97.3%**	637	99.2%	**A**	-	**B**	-	**N370S**
1	156,456,301	rs202206511	-1,250,667	**AB**	**BB**	2277	**98.2%**	1721	**98.2%**	**A**	-	**B**	-	**N370S**
1	156,466,699	rs1342442	-1,261,065	**AB**	**BB**	970	**96.2%**	767	99.1%	**A**	-	**B**	-	**N370S**
1	156,610,688	rs386529395	-1,405,054	**AB**	**BB**	1134	**98.1%**	1048	99.6%	**A**	-	**B**	-	**N370S**
1	156,907,550	rs3765790	-1,701,916	**AB**	**AA**	7874	**2.5%**	7854	0.4%	**B**	-	**A**	-	**N370S**
1	156,908,986	rs386585816	-1,703,352	**AB**	**AA**	2069	**1.6%**	2007	**1.3%**	**B**	-	**A**	-	**N370S**
1	156,933,962	rs11806313	-1,728,328	**AB**	**AA**	5269	**1.4%**	5923	**2.2%**	**B**	-	**A**	-	**N370S**
1	156,964,467	rs6689412	-1,758,833	**AB**	**AA**	7817	0.6%	7865	**1.5%**	**B**	-	**A**	-	**N370S**
1	156,976,225	rs822581	-1,770,591	**AB**	**BB**	1634	**97.9%**	1062	99.4%	**A**	-	**B**	-	**N370S**
1	156,977,905	rs703152	-1,772,271	**AB**	**BB**	7869	**95.9%**	7874	99.6%	**A**	-	**B**	-	**N370S**
1	157,059,785	rs2031626	-1,854,151	**AB**	**AA**	620	**1.5%**	883	**1.4%**	B	-	-	-	-
1	157,061,220	rs6677868	-1,855,586	**AB**	**AA**	7744	0.9%	7794	**2.4%**	B	-	-	-	-

1	157,090,136	rs12022068	-1,884,502	**AB**	**AA**	7932	**2.7%**	7932	**1.7%**	**B**	-	**A**	-	**N370S**
1	157,204,044	rs12039209	-1,998,410	**AB**	**AA**	2073	**2.4%**	1412	0.8%	B	-	-	-	-

Abbreviations and footnotes are the same as in Supplemental Table 6 with the following footnote and abbreviation modifications:

E. The paternal fetal haplotype (PHiF) was determined from SNP III data (as described in Supplemental Methods); the paternal N370S-linked (PFB N370S) and paternal del55-linked (PFB del55) haplotypes were determined by family-based linkage analysis; the N370S consensus haplotype (N370S cons) was derived according to Figure 2. An "-" indicates that no haplotype data was available at the given position. Bold alleles were used for diagnosis of the paternal allele in the fetus ("DPAiF").

F. Fetal allele identity (FAI) was determined by comparing the "PHiF" haplotype to the "PFB N370S", "PFB del55", and/or "N370S cons" haplotypes

G. Near consensus N370S haplotype as determined according to Figure 4.

Supplemental Table 19. Identification of the paternal allele in the family 6 fetus (large panel)

Variant data							plasma DNA rep 6-1		plasma DNA rep 6-2		Haplotypes[E]			FAI[F]
Ch	Position (hg19)	dbSNP ID[A]	DFM (bps)	PGT[B]	MGT[B]	FL[C] (%)	RD (x)	BAF[D]	RD (x)	BAF[D]	PHiF	PFB N370S	N370S cons	DPAiF
1	153,310,156	rs3006444	1,895,478	**AB**	**BB**		445	99.1%	404	**97.5%**	A	-	-	-
1	153,776,666	rs4341393	1,428,968	**AB**	**BB**		499	**98.0%**	1287	**93.4%**	**A**	**A**	-	**N370S**
1	153,794,996	rs6427298	1,410,638	**AB**	**BB**		140	**95.7%**	111	100.0%	A	-	-	-
1	153,813,165	rs4370789	1,392,469	**AB**	**BB**		110	100.0%	237	**98.7%**	**A**	**A**	-	**N370S**
1	154,017,501	rs2841102	1,188,133	**AB**	**AA**		3615	**2.8%**	4151	**2.4%**	**B**	**B**	-	**N370S**
1	154,026,642	rs34752019	1,178,992	**AB**	**AA**		6041	**1.6%**	3044	1.0%	B	-	-	-
1	154,556,663	rs1127311	648,971	**AB**	**BB**	3.6	393	**98.0%**	357	100.0%	**A**	**A**	-	**N370S**
1	154,589,965	rs9426827	615,669	**AB**	**BB**		246	100.0%	292	**94.5%**	**A**	**A**	-	**N370S**
1	154,609,009	rs9427109	596,625	**AB**	**BB**		1938	**83.7%**	1062	**98.1%**	**A**	**A**	-	**N370S**
1	154,653,951	rs61811421	551,683	**AB**	**AA**		555	0.2%	827	**1.5%**	B	-	-	-
1	154,677,716	rs56320421	527,918	**AB**	**AA**		7922	**8.7%**	7881	0.4%	B	-	-	-
1	154,774,966	rs6690891	430,668	**AB**	**AA**		2549	**2.0%**	3724	1.4%	**B**	**B**	-	**N370S**
1	155,030,557	rs11264303	175,077	**AB**	**BB**		1891	**96.4%**	871	99.3%	**A**	**A**	-	**N370S**

1	155,205,634	**N370S**	-	**-**	**-**	-	-	-	-	-	-	-	-
1	155,213,124	rs11264345	-7,490	**AB**	**BB**	7672	**97.8%**	7692	99.4%	**A**	**A**	**A**	**N370S**
1	155,230,131	rs386580544	-24,497	**AB**	**BB**	4312	**96.4%**	7656	**97.4%**	**A**	**A**	**A**	**N370S**
1	155,473,356	rs1360554	-267,722	**AB**	**AA**	874	**6.5%**	2205	1.0%	**B**	**B**	**B**	**N370S**
1	155,893,750	rs506688	-688,116	**AB**	**BB**	335	100.0%	424	**98.3%**	**A**	**A**	**A**[G]	**N370S**
1	156,106,863	rs553016	-901,229	**AB**	**AA**	140	**5.0%**	159	1.3%	**B**	-	**B**[G]	**N370S**
1	156,502,991	rs1609666	-1,297,357	**AB**	**BB**	1928	**98.2%**	1567	**95.3%**	**A**	**A**	-	**N370S**
1	156,532,733	rs4661183	-1,327,099	**AB**	**AA**	7731	**3.5%**	7645	0.8%	**B**	**B**	-	**N370S**
1	156,588,749	rs2365714	-1,383,115	**AB**	**BB**	3951	**70.6%**	2353	**98.2%**	**A**	**A**	-	**N370S**
1	156,589,450	rs3795727	-1,383,816	**AB**	**BB**	336	**95.2%**	276	99.3%	**A**	**A**	-	**N370S**
1	156,711,623	rs3806417	-1,505,989	**AB**	**AA**	532	**36.8%**	1020	0.2%	B	-	-	-
1	156,814,027	rs56252149	-1,608,393	**AB**	**AA**	482	1.0%	323	**19.2%**	B	-	-	-
1	157,043,969	rs16865471	-1,838,335	**AB**	**AA**	262	**2.3%**	1129	1.3%	B	-	-	-
1	157,194,059	rs10494311	-1,988,425	**AB**	**BB**	855	**95.3%**	816	99.3%	**A**	**A**	-	**N370S**

Abbreviations and footnotes are the same as in Supplemental Table 6 with the following additional footnote:

G. Near consensus N370S haplotype as determined according to Figure 4.

Supplemental Table 20. Identification of the maternal allele in the family 7 fetus (large panel)

Variant data							plasma DNA rep 7-1		plasma DNA rep 7-2		Haplotypes[E]				FAI[F]
Ch	Position (hg19)	dbSNP ID[A]	DFM (bps)	PGT[B]	MGT[B]	FL[C] (%)	RD (x)	BAF[D]	RD (x)	BAF[D]	MHiF	MFB N370S	MFB L444P	N370S cons	DMAiF
1	153,217,977	rs1926234	1,987,657	**BB**	**AB**		29461	67.3%	29390	74.6%	**B**	**A**	**B**	-	**L444P**
1	153,335,384	rs386537988	1,870,250	**AA**	**AB**		6853	18.8%	8928	21.4%	A	-	-	-	-
1	153,526,349	rs55955983	1,679,285	**AA**	**AB**		12549	32.5%	33520	33.4%	**A**	-	**A**	-	**L444P**
1	153,776,666	rs4341393	1,428,968	**AA**	**AB**		9602	27.8%	11192	43.6%	A	-	-	-	-
1	153,777,848	rs1139620	1,427,786	**AA**	**AB**		1767	26.3%	641	16.2%	A	-	-	-	-
1	153,779,412	rs1127091	1,426,222	**AA**	**AB**		15005	29.0%	9780	32.6%	A	-	-	-	-
1	153,825,208	rs4540690	1,380,426	**AA**	**AB**	23.2	8827	44.8%	8411	36.7%	A	-	-	-	-
1	153,854,161	rs12039936	1,351,473	**AA**	**AB**		538	30.7%	763	30.3%	**A**	-	**A**	-	**L444P**
1	153,893,023	rs10494303	1,312,611	**BB**	**AB**		8757	54.7%	7414	54.6%	B	-	-	-	-
1	154,167,788	rs4364874	1,037,846	**BB**	**AB**		10931	70.1%	12874	56.2%	B	-	-	-	-
1	154,192,143	rs9803862	1,013,491	**BB**	**AB**		2507	69.2%	2222	53.1%	B	-	-	-	-
1	154,741,579	rs7547552	464,055	**AA**	**AB**		7330	38.0%	8184	24.6%	A	-	-	-	-
1	154,774,966	rs6690891	430,668	**AA**	**AB**		4793	27.3%	4395	30.8%	**A**	-	**A**	-	**L444P**

1	154,803,645	rs386523166	401,989	**BB**	**AB**	8516	55.2%	6881	57.4%	**B**	-	**B**	-	**L444P**
1	154,810,548	rs34647098	395,086	**AA**	**AB**	15074	38.9%	14238	39.4%	**A**	-	**A**	-	**L444P**
1	154,828,683	rs56382016	376,951	**AA**	**AB**	1421	18.2%	2680	28.1%	**A**	-	**A**	-	**L444P**
1	155,205,634	**N370S**	-	**-**	**-**	-	-	-	-	-	-	-	-	-
1	156,351,699	rs3748569	-1,146,065	**AA**	**AB**	1450	40.1%	2048	34.9%	**A**	-	**A**	-	**L444P**
1	156,351,792	rs386585381	-1,146,158	**AA**	**AB**	2749	37.5%	3902	33.1%	**A**	-	**A**	-	**L444P**
1	156,718,448	rs11264533	-1,512,814	**AA**	**AB**	10379	30.0%	9858	29.8%	A	-	-	-	-
1	156,738,464	rs76807267	-1,532,830	**AA**	**AB**	1617	25.4%	3266	38.3%	**A**	-	**A**	-	**L444P**
1	156,774,672	rs2644609	-1,569,038	**AA**	**AB**	1634	35.6%	2430	33.3%	**A**	**B**	**A**	-	**L444P**
1	156,805,803	rs7534418	-1,600,169	**BB**	**AB**	185	79.5%	368	98.9%	**B**	**A**	**B**	-	**L444P**
1	156,855,854	rs61813800	-1,650,220	**AA**	**AB**	654	16.2%	1804	12.6%	**A**	-	**A**	-	**L444P**
1	156,934,893	rs4661077	-1,729,259	**AA**	**AB**	2413	29.8%	1735	35.3%	**A**	**B**	**A**	-	**L444P**

Abbreviations and footnotes are the same as in Supplemental Table 13 with the following footnote and abbreviation modifications:

E. The maternal fetal haplotype (MHiF) was determined from SNP III data (as described in Supplemental Methods); the maternal N370S-linked (MFB N370S) and maternal L444P-linked (MFB L444P) haplotypes were determined by family-based linkage analysis; the N370S consensus haplotype (N370S cons) was derived according to Figure 2. An "-" indicates that no haplotype data was available at the given position. Bold alleles were used for diagnosis of the maternal allele in the fetus ("DMAiF").

F. Fetal allele identity (FAI) was determined by comparing the "MHiF" haplotype to the "MFB N370S", "MFB L444P", and/or "N370S cons" haplotypes

Supplemental Table 21. Identification of the maternal allele in the family 8 fetus (large panel)

Variant data							plasma DNA rep 8-1		plasma DNA rep 8-2		Haplotypes[E]				FAI[F]
Ch	Position (hg19)	dbSNP ID[A]	DFM (bps)	PGT[B]	MGT[B]	FL[C] (%)	RD (x)	BAF[D]	RD (x)	BAF[D]	MHiF	MFB N370S	MFB 84GG	N370S cons	DMAiF
1	153,283,978	rs821420	1,921,656	**BB**	**AB**		4411	62.3%	2660	52.0%	**B**	**A**	-	-	**84GG**
1	153,284,423	rs821418	1,921,211	**BB**	**AB**		1917	84.0%	1176	73.0%	**B**	**A**	-	-	**84GG**
1	153,302,417	rs2771120	1,903,217	**BB**	**AB**		820	78.3%	1131	88.8%	**B**	**A**	-	-	**84GG**
1	153,327,732	rs2916198	1,877,902	**AA**	**AB**		694	20.3%	209	24.9%	**A**	-	**A**	-	**84GG**
1	153,609,039	rs4845557	1,596,595	**AA**	**AB**		538	5.2%	864	25.3%	A	-	-	-	-
1	153,613,545	rs12564925	1,592,089	**AA**	**AB**		802	3.9%	1323	13.2%	A	-	-	-	-
1	153,616,857	rs3790412	1,588,777	**AA**	**AB**	2.8	7859	40.8%	3529	24.0%	A	-	-	-	-
1	153,753,079	rs12030242	1,452,555	**AA**	**AB**		7847	5.8%	7707	35.9%	**A**		**A**	-	**84GG**
1	154,238,383	rs1194596	967,251	**AA**	**AB**		3814	31.2%	3757	21.1%	**A**	**B**	**A**	-	**84GG**
1	154,259,275	rs1760802	946,359	**AA**	**AB**		942	5.5%	2605	2.2%	**A**	**B**	**A**	-	**84GG**
1	154,381,103	rs12083537	824,531	**AA**	**AB**		1506	41.4%	798	28.3%	A		-	-	-
1	154,692,088	rs4845663	513,546	**BB**	**AB**		7861	69.5%	7876	56.7%	**B**	**A**	-	-	**84GG**
1	154,731,151	rs2131900	474,483	**AA**	**AB**		7146	12.9%	5791	36.9%	**A**	**B**	-	-	**84GG**

1	154,737,423	rs1506983	468,211	**AA**	**AB**	912	29.1%	1091	7.5%	**A**	**B**	-	-	**84GG**
1	154,741,357	rs56033137	464,277	**AA**	**AB**	145	40.7%	268	20.5%	**A**	**B**	-	-	**84GG**
1	154,811,677	rs1218574	393,957	**BB**	**AB**	1887	82.6%	2373	61.9%	**B**	-	**B**	-	**84GG**
1	155,205,634	**N370S**	-	**-**	**-**	-	-	-	-	-	-	-	-	-
1	155,870,416	rs1749409	-664,782	**BB**	**AB**	1072	63.0%	1392	66.6%	**B**	-	**B**	**A**^{G}	**84GG**
1	155,899,020	rs539602	-693,386	**BB**	**AB**	406	51.5%	783	57.1%	**B**	-	**B**	**A**^{G}	**84GG**
1	155,907,823	rs11264422	-702,189	**BB**	**AB**	4830	57.8%	7898	56.7%	**B**	-	**B**	**A**^{G}	**84GG**
1	156,045,662	rs10047112	-840,028	**BB**	**AB**	333	62.8%	321	75.4%	**B**	**A**		**A**^{G}	**84GG**
1	156,089,920	rs610918	-884,286	**AA**	**AB**	7797	24.0%	6685	7.4%	**A**	-	**A**	**B**^{G}	**84GG**
1	156,099,669	rs513043	-894,035	**AA**	**AB**	1225	14.7%	231	13.4%	**A**	-	**A**	**B**^{G}	**84GG**
1	156,337,134	rs3806410	-1,131,500	**AA**	**AB**	5764	16.9%	2573	25.0%	**A**	-	**A**	-	**84GG**
1	156,645,948	rs77658164	-1,440,314	**AA**	**AB**	943	47.8%	240	33.8%	A	-	-	-	-
1	156,976,393	rs863717	-1,770,759	**AA**	**AB**	4232	19.0%	2548	32.5%	A	-	-	-	-

Abbreviations and footnotes are the same as in Supplemental Table 13 with the following footnote and abbreviation modifications:

E. The maternal fetal haplotype (MHiF) was determined from SNP III data (as described in Supplemental Methods); the maternal N370S-linked (MFB N370S) and maternal 84GG-linked (MFB 84GG) haplotypes were determined by family-based linkage analysis; the N370S consensus haplotype (N370S cons) was derived according to Figure 2. An "-" indicates that no haplotype data was available at the given position. Bold alleles were used for diagnosis of the maternal allele in the fetus ("DMAiF").

F. Fetal allele identity (FAI) was determined by comparing the "MHiF" haplotype to the "MFB N370S", "MFB 84GG", and/or "N370S cons" haplotypes