Supplemental data

Supplemental Table 1 List of diseases for which prenatal diagnosis was performed.

Disease	OMIM	Inheritance	Incidence	Gene	OMIM	Exon	CV	Amnio
Myotonic dystrophy type I[a]	160900	AD	1:8.000	*DMPK*	605377	15	10	5
Huntington's disease[a]	143100	AD	1:25.000	*HTT*	613004	67	10	0
Tuberous sclerosis	191100	AD	1:10.000	*TSC1*	605284	23	8	1
				TSC2	191092	41		
Achondroplasia[a]	100800	AD	1:25.000	*FGFR3*	134934	17	0	8
Autosomal dominant deafness 3A[a]	601544	AD/AR	1:35.000	*GJB2*	121011	2	3	0
Spinocerebellar ataxia 3[a]	109150	AD	1:30.000	*ATXN3*	607047	11	2	0
Multiple Endocrine Neoplasia Type IIA[a]	171400	AD	1:50.000	*RET*	164761	20	2	0
Holt-Oram syndrome	142900	AD	1:100.000	*TBX5*	601620	10	0	2
Spinocerebellar ataxia 2[a]	183090	AD	1:50.000	*ATXN2*	601517	25	1	1
Facioscapulohumeral muscular dystrophy 1A	158900	AD	1:15.000				1	0
Spinocerebellar ataxia 1[a]	164400	AD	1:100.000	*ATXN1*	601556	9	1	0
Familial Adenomatous Polyposis 1[a]	175100	AD	1:20.000	*APC*	611731	18	1	0
Von Hippel-Lindau Disease	193300	AD	1:36.000	*VHL*	193300	3	1	0
Multiple Endocrine Neoplasia Type I[a]	131100	AD	1:100.000	*MEN1*	613733	10	0	1
Autoimmune polyglandular syndrome type I	240300	AR		*AIRE*	607358	14	1	0
Argininosuccinic aciduria[a]	207900	AR	1:70.000	*ASA lyase*	608310	17	0	5
Friedreich's ataxia	293000	AR	1:50.000	*FRDA*	606829	7	1	0
Carbamoyl phosphate synthetase 1 deficiency[a]	237300	AR	1:500.000	*CPSI*	608307	38	1	0
Neuronal Ceroid Lipofuscinosis	256730	AR	1:20.000	*PPT1*	600722	9	1	0
Congenital 21-Hydroxylase Deficiency[a]	201910	AR	1:15.000	*CYP21A2*	613815	10	11	0
Carbonic Anhydrase II Deficiency	259730	AR	1:50.000	*CA*	611492	7	2	0
Congenital Disorders of Glycosylation 1a	212065	AR	1:20.000	*PMM2*	601785	8	1	0
Pyruvate Carboxylase Deficiency	266150	AR	1:250.000	*PC*	608786	19	1	0
Pyruvate Kinase Deficiency	266200	AR	1:20.000	*PKLR*	609712	12	3	0
Phenylketonuria[a]	261600	AR	1:15.000	*PAH*	612439	13	4	6
Cystic Fibrosis[a]	219700	AR	1:2.500	*CFTR*	602421	27	167	14
Galactosaemia	230400	AR	1:50.000	*GALT*	606999	11	1	2
Glycogen Storage Disease Type Ib	232220	AR	1:100.000	*SLC37A4*	602671	9	0	1
Glycogen Storage Disease Type II	232300	AR	1:100.000	*GAA*	606800	20	10	0
Glycogen Storage Disease Type III	232400	AR	1:100.000	*AGL*	610860	35	1	0
Glycogen Storage Disease Type IV	232500	AR	<1:100.000	*GBE1*	607839	16	1	1
GM1-Gangliosidosis Type I	230500	AR	1:150.000	*GLB1*	611458	16	4	1
GM2 Gangliosidosis Type II	268800	AR	<1:100.000	*HEXB*	606873	14	1	0
Lysinuric Protein Intolerance[a]	222700	AR	<1:100.000	*SLC7A7*	603593	11	1	0
Glycine encephalopathy	605899	AR	1:60.000	*AMT*	238310	9	3	0
				GLDC	238300	25		
				GCSH	238330	5		
Maple syrup urine disease type II	248600	AR	1:180.000			1	0	
Krabbe Leukodystrophy	245200	AR	1:100.000	*GALC*	606890	17	7	1
Methylmalonic Aciduria[a]	251000	AR	1:80.000	*MUT*	609058	3	6	6
Propionic Acidaemia[a]	606054	AR	1:35.000	*PCCB*	232050	15	0	2
Molybdenum cofactor deficiency	252150	AR	<1:100.000	*MOCS2*	603708	7	1	0
				MOCS1	603707	10		
				GEPH	603930	22		
Mucolipidosis III gamma	252605	AR	*GNPTG*	607838	11	0	2	
Mucopolysaccharidosis Type I[a]	607014	AR	1:100.000	*IDUA*	252800	14	2	5
Mucopolysaccharidosis Type IIIA[a]	252900	AR	1:100.000	*SGSH*	605270	8	3	2
Mucopolysaccharidosis Type IIIB[a]	252920	AR	1:100.000	*NAGLU*	609701	6	0	4
Niemann-Pick Disease type A	257200	AR	1:250.000	*SMPD1*	607608	6	9	2
Non-ketotic hyperglycinaemia	605899	AR	<1:100.000	*GLDC*	238200	25	1	0
Smith-Lemli-Opitz Syndrome	270400	AR	1:30.000	*DHCR7*	602858	7	6	3

(**Supplemental Table 1** Continued)

Disease	OMIM	Inheritance	Incidence	Gene	OMIM	Exon	CV	Amnio
Zellweger Syndrome	214100	AR	1:40.000	*PEX1*	602136	24	2	0
Canavan Disease	271900	AR	<1:100.000	*ASPA*	608034	6	1	4
Shwachman Diamond Syndrome[a]	260400	AR	1:50.000	*SBDS*	607444	5	1	0
HHH Syndrome	238970	AR	<1:100.000	*SLC25A15*	603861	7	1	0
Spinal Muscular Atrophy[a]	AR		1:10.000	*SMN1*	600354	9	33	21
Angelman Syndrome	105830	AR	1:20.000	*UBE3A*	601623		2	4
β-Thalassaemia[a]	613985	AR		*HBB*	141900	3	405	31
α-Thalassaemia[a]	604131	AR		*MBA1*	141800	3	6	1
Haemoglobin variant[a]		AR		*HBB*	141900	3	33	0
β-thalassaemia/haemoglobin variant[a]							38	3
δ-β-thalassaemia/β thalassaemia[a]							4	2
Fanconi anaemia	227650	AR	1:350.000				5	1
Choroideraemia[a]	303100	XR	1:50.000[a]	*CHM*	300390	15	2	0
Ornithine Transcarbamylase Deficiency[a]	311250	XR	1:20.000	*OTC*	300461	10	3	1
Duchenne/Becker Muscular Dystrophy[a]	310200	XR	1:3.000	*DMD*	300377	79	77	6
Haemophilia A[a]	306700	XR	1:5.000	*F8*	300841	26	31	5
Haemophilia B[a]	306900	XR	1:30.000	*F9*	306900	8	5	3
X-Linked Hydrocephalus	307000	XR	1:30.000	*L1CAM*	308840	28	2	0
Lesch-Nyhan syndrome	300322	XR	1:380.000	*HPRT1*	308000	9	4	0
Mucopolysaccharidosis Type II[a]	309900	XR	1:250.000	*IDS*	300823	9	15	4
Rett Syndrome	312750	XR	1:15.000[b]	*MECP2*	300005	3	2	1
Severe Combined Immunodeficiency[a]	300400	XR	1:100.000	*IL2RG*	308380	8	5	0
Wiskott Aldrich Syndrome	301000	XR	1:250.000	*WAS*	300392	12	1	0
Fragile X mental retardation syndrome[a]	300624	XR	1:3600	*FMR1*	309950	17	24	14
X-Linked Mental Retardation	300419	XR		*ARX*	300382	5	1	0
X-linked Centronuclear Myopathy	310400	XR	1:5000	*MTM1*	300415	15	2	0

AD, autosomal dominant; amnio, amniocentesis; AR, autosomal recessive; CV, chorionic villi sampling; XR, X-linked recessive.
[a]Prenatal diagnoses performed in our laboratory. [b]The incidence of X-linked recessive diseases refers to male newborns, with the exception of Rett syndrome which occurs only in females. The incidence of some diseases is not indicated because it differs greatly among ethnic groups and geographic areas.