

Gen_Genetic_Test

Genetic Test

Subjectkey (NDAR GUID)	
Source subject ID	
Age (in months)	
Genetic test	
Date of test	
Genetic test results	Normal Abnormal
Comments	
Test provider	

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Example of table for NDAR data submission:

Subjectkey	Src_subject_id	Interview_age	Interview_date	Genetic_test	Test_result	Test_provider	Genetic_test_notes
NDAR_INVEW671NRA	MT_R21_006	36	1/1/2008	ASD_NRXN1	Normal	GeneDx	none
NDAR_INVEW671NRA	MT_R21_006	36	1/1/2008	NF2_NF2	Abnormal	GeneDx	none

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#	Test	Marker	Chromosome	Chromosome Location	Condition	Gene Name
1	Test1_1p32-p31	1p32-p31	1	1p32-p31	Chromosome 1p32-p31 deletion syndrome	NFIA
2	Test2_SLC2A1	SLC2A1	1	1p34.2	GLUT1 deficiency syndrome type 1 and type 2	GLUT1
3	Test3_1p36	1p36	1	1p36	Chromosome 1p36 deletion syndrome	
4	Test4_1q21.1	1q21.1	1	1q21.1	Chromosome 1q21.1 deletion syndrome	
5	Test5_1q41q42	1q41q42	1	1q41-q42	1q41-q42 deletion syndrome	DISP1, SUSD4, CAPN2, TP53BP2, FBXO28
6	Test6_2p16.1-p15	2p16.1-p15	2	2p16.1-p15	Chromosome 2p16.1-p15 deletion syndrome	
7	Test7_NRXN1	NRXN1	2	2p16.3	Autism Spectrum Disorders	NRXN1
8	Test8_SOS1	SOS1	2	2p22.1	Noonan syndrome 4	SOS1
9	Test9_2q13	2q13	2	2q13	Autism Spectrum Disorders	
10	Test10_NPHP1	NPHP1	2	2q13	Joubert syndrome	NPHP1
11	Test11_ZEB2	ZEB2	2	2q22	Mowat-Wilson syndrome	ZEB2
12	Test12_MBD5	MBD5	2	2q23.1	Rett syndrome	MBD5
13	Test13_SLC4A10	SLC4A10	2	2q23-q24	Autism Spectrum Disorders	SLC4A10
14	Test14_SCN1A	SCN1A	2	2q24	Generalized epilepsy with febrile seizures (GEFS+)	SCN1A
15	Test15_2q33.3-q35	2q33.3-q35	2	2q33.3-q35	Developmental delay	
16	Test16_HDAC4	HDAC4	2	2q37.3	Brachydactyly-mental retardation syndrome (BDRM)	HDAC4
17	Test17_FOXP1	FOXP1	3	3p13	Mental retardation with language impairment and autistic features	FOXP1
18	Test18_RAF1	RAF1	3	3p25	Noonan syndrome, Leopard syndrome	RAF1
19	Test19_CNTN4	CNTN4	3	3p26.3	Autism Spectrum Disorders	CNTN4
20	Test20_ARL13B	ARL13B	3	3q11.2	Joubert syndrome	ARL13B

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21	Test21_3q29	3q29	3	3q29	3q29 microdeletion syndrome, 3q29 microduplication syndrome	
22	Test22_CC2D2A	CC2D2A	4	4p15.33	Joubert syndrome	CC2D2A
23	Test23_4p16.3	4p16.3	4	4p16.3	Wolf-Hirschhorn syndrome	WHSC1
24	Test24_4q32-q33	4q32-q33	4	4q32-q33	Learning difficulties, autistic features	
25	Test25_4q35	4q35	4	4q35	Fascio-scapulo-humeral muscular dystrophy (FSHD)	
26	Test26_NIPBL	NIPBL	5	5p13.2	Cornelia de Lange syndrome (CdLS)	NIPBL
27	Test27_5p15.2	5p15.2	5	5p15.2	Cri du Chat syndrome	TERT, CTNND2,SEMAF
28	Test28_MEF2C	MEF2C	5	5q14.3	5q14 microdeletion syndrome	MEF2C
29	Test29_NSD1	NSD1	5	5q35	Sotos syndrome	NSD1
30	Test30_SYNGAP1	SYNGAP1	6	6p21.32	Autosomal Dominant Mental Retardation 5	SYNGAP1
31	Test31_GRIK2	GRIK2	6	6q16.3-q21	Autosomal Recessive Mental Retardation	GRIK2
32	Test32_SIM1	SIM1	6	6q16.3-q21	Prader-Willi syndrome	SIM1
33	Test33_AHI1	AHI1	6	6q23.3	Joubert syndrome	AHI1
34	Test34_6q25.1-q27	6q25.1-q27	6	6q25.1-q27	Physical features, hearing loss, developmental delay, and varying degrees of intellectual disability	TIAM2, NOX3, SYNJ2
35	Test35_AUTS2	AUTS2	7	7q11.22	Autism Spectrum Disorders	AUTS2
36	Test36_ELN	ELN	7	7q11.23	Williams syndrome	ELN
37	Test37_RELN	RELN	7	7q22	Norman-Roberts syndrome	RELN
38	Test38_FOXP2	FOXP2	7	7q31	developmental verbal dyspraxia	FOXP2
39	Test39_BRAF	BRAF	7	7q34	Noonan syndrome, Costello syndrome, cardiofaciocutaneous syndrome (CFC syndrome), Leopard syndrome	BRAF
40	Test40_CNTNAP2	CNTNAP2	7	7q35	Autism Spectrum Disorders	ASD
41	Test41_HGSNAT	HGSNAT	8	8p11.21	Sanfilippo syndrome	HGSNAT
42	Test42_8q11.23	8q11.23	8	8q11.23	Autism Spectrum Disorders	
43	Test43_CHD7	CHD7	8	8q12.1	CHARGE syndrome	CHD7
44	Test44_TM67	TM67	8	8q22.1	Joubert syndrome	TM67/MKS3

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45	Test45_COH1	COH1	8	8q22-q23	Cohen syndrome	COH1
46	Test46_KCNQ3	KCNQ3	8	8q24	idiopathic epilepsies	KCNQ3
47	Test47_8q24.11-q24.13	8q24.11-q24.13	8	8q24.11-q24.13	Langer-Giedion syndrome	TRPS1, EXT1
48	Test48_9q22.3	9q22.3	9	9q22.3	Gorlin syndrome	PTCH1
49	Test49_TSC1	TSC1	9	9q34	Tuberous sclerosis complex	TSC1
50	Test50_EHMT1	EHMT1	9	9q34.3	Kleefstra syndrome	EHMT1
51	Test51_INPP5E	INPP5E	9	9q34.3	Joubert syndrome	INPP5E
52	Test52_DGS2	DGS2	10	10p15.1-p14	DiGeorge syndrome	DGS2
53	Test53_10q22.3-23.2	10q22.3-23.2	10	10q22.3-23.31	Banyan-Riley-Ruvalcaba syndrome, Cowden syndrome	PTEN, BMPR1A
54	Test54_PTEN	PTEN	10	10q23.3	Bannayan-Riley-Ruvalcaba syndrome	PTEN
55	Test55_SHOC2	SHOC2	10	10q25.2	Noonan syndrome	SHOC2
56	Test56_SMC3	SMC3	10	10q25.2	Cornelia de Lang syndrome 3	SMC3
57	Test57_10q26	10q26	10	10q26	10q deletion syndrome	HMX2, HMX3
58	Test58_11p11.2	11p11.2	11	11p11.2	Potocki-Shaffer syndrome	ALX4
59	Test59_WT1	WT1	11	11p13	WAGR syndrome	WT1
60	Test60_HRAS	HRAS	11	11p15.5	Costello syndrome, Noonan syndrome, and cardiofaciocutaneous syndrome (CFC syndrome)	HRAS
61	Test61_TMEM216	TMEM216	11	11q12.2	Joubert syndrome	TMEM216
62	Test62_SHANK2	SHANK2	11	11q13.3-q13.4	Autism Spectrum Disorders	SHANK2
63	Test63_DHCR7	DHCR7	11	11q13.4	Smith-Lemli-Opitz syndrome	DHCR7
64	Test64_11q23-qter	11q23-qter	11	11q23-qter	Jacobsen syndrome	BSX, FLI-1, ETS-1, JAM3
65	Test65_KRAS	KRAS	12	12p12.1	Costello syndrome, Noonan syndrome, and cardiofaciocutaneous syndrome (CFC syndrome)	KRAS
66	Test66_CACNA1C	CACNA1C	12	12p13.3	Timothy syndrome	CACNA1C
67	Test67_12q13.3-q15	12q13.3-q15	12	12q13.3-q15	Silver-Russell syndrome	HMGA2, LEMD3
68	Test68_GNS	GNS	12	12q14	Sanfilippo syndrome, also called mucopolysaccharidosis (MPS) type III	GNS

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69	Test69_TPH2	TPH2	12	12q21.1	Tourett syndrome	TPH2
70	Test70_CEP290	CEP290	12	12q21.32	Joubert syndrome	CEP290
71	Test71_PTPN11	PTPN11	12	12q24.1	Noonan syndrome	PTPN11
72	Test72_Trisonomy 13	Trisonomy 13	13	13	Trisonomy 13, also known as Patau syndrome	
73	Test73_13q	13q	13	13q11	13q deletion syndrome, 13q duplication syndrome (also called partial trisomy 13)	RB1
74	Test74_PCDH9	PCDH9	13	13q21.32	Autism Spectrum Disorders	PCDH9
75	Test75_FOXG1	FOXG1	14	14q13	Rett syndrome	FOXG1
76	Test76_14q23.2-q23.3	14q23.2-q23.3	14	14q23.2-q23.3	Spherocytosis and intellectual disability	SPTB
77	Test77_SNORD115-1	SNORD115-1	15	15q11.2	Prader-Willi syndrome	SNORD115-1
78	Test78_GABRB3	GABRB3	15	15q11.2-q12	Autism Spectrum Disorders	GABRB3
79	Test79_15q11-q12	15q11-q12	15	15q11-q12	Angelman syndrome, Prader-Willi syndrome (PWS), Chromosome 15q duplication syndrome	UBE3A
80	Test80_15q13	15q13	15	15q13	15q13.3 microdeletion syndrome, duplication 15q	
81	Test81_MAP2K1	MAP2K1	15	15q21	Cardiofaciocutaneous syndrome	MAP2K1
82	Test82_GATM	GATM	15	15q21.1	Severe cognitive impairment and global developmental delay	GATM
83	Test83_15q24.1-q24.2	15q24.1-q24.2	15	15q24.1-q24.2	15q24.1 deletion syndrome	
84	Test84_16p11.2	16p11.2	16	16p11.2	16p11.2 microdeletion syndrome, 16p11.2 microduplication syndrome	
85	Test85_16p13.1	16p13.1	16	16p13.1	Autism Spectrum Disorders	
86	Test86_16p13.3	16p13.3	16	16p13.3	Alpha-thalassemia, cognitive impairment	ATR-16, SOX8
87	Test87_A2BP1	A2BP1	16	16p13.3	Autism Spectrum Disorders	A2BP1
88	Test88_CREBBP	CREBBP	16	16p13.3	Rubenstein-Taybi syndrome (RTS)	CREBBP
89	Test89_TSC2	TSC2	16	16p13.3	Tuberous sclerosis complex	TSC2
90	Test90_RPGRIP1L	RPGRIP1L	16	16q12.2	Joubert syndrome	RPGRIP1L
91	Test91_17p11.2	17p11.2	17	17p11.2	Smith-Magenis syndrome, Potocki-Lupski syndrome	RAI1

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92	Test92_17p13.3	17p13.3	17	17p13.3	Miller-Dieker syndrome, also known as Lissencephaly type 1	LIS1, YWHAE
93	Test93_NF1	NF1	17	17q11.2	Neurofibromatosis type I	NF1
94	Test94_17q12	17q12	17	17q12	Autism Spectrum Disorders	HNF1B gene
95	Test95_NAGLU	NAGLU	17	17q21.2	Sanfilippo syndrome	NALGU
96	Test96_17q21.31	17q21.31	17	17q21.31	17q21.31 microduplication syndrome	
97	Test97_SGSH	SGSH	17	17q25.3	Sanfilippo syndrome	SGSH
98	Test98_Trisonomy 18	Trisonomy 18	18	18	Trisonomy 18, also known as Edwards syndrome	
99	Test99_TCF4	TCF4	18	18q21.2	Pitt-Hopkins syndrome	TCF4
100	Test100_GAMT	GAMT	19	19p13.3	Severe developmental delay	GAMT
101	Test101_MAP2K2	MAP2K2	19	19p13.3	Noonan syndrome, Costello syndrome, and cardiofaciocutaneous syndrome (CFC syndrome)	MAP2K2
102	Test102_Trisonomy 21	Trisonomy 21	21	21	Down syndrome	
103	Test103_22q11.2	22q11.2	22	22q11.2	DiGeorge syndrome	TBX1
104	Test104_NF2	NF2	22	22q12.2	Neurofibromatosis type II	NF2
105	Test105_ADSL	ADSL	22	22q13.1	ADSL deficiency	ADSL
106	Test106_SOX10	SOX10	22	22q13.1	Waardenburg-Shah syndrome type 2E (WS2E) and type 4C (WS4C), as well as PCWH (Peripheral demyelinating neuropathy, Central dysmyelination, Waardenbug syndrome, and Hirschsprung disease)	SOX10
107	Test107_EP300	EP300	22	22q13.2	Rubenstein-Taybi syndrome (RTS)	EP300
108	Test108_SHANK3	SHANK3	22	22q13.31-q13.33	Autism Spectrum Disorders	SHANK3
109	Test109_XXY	XXY	X	X	Klinefelter syndrome (47,XXY)	
110	Test110_FGD1	FGD1	X	Xp11.21	X-linked mental retardation	FGD1
111	Test111_HUWE1	HUWE1	X	Xp11.22	X-linked mental retardation	HUWE1
112	Test112_PHF8	PHF8	X	Xp11.22	X-linked mental retardation	PHF8
113	Test113_SHROOM4	SHROOM4	X	Xp11.22	X-linked mental retardation	SHROOM4
114	Test114_KDM5C	KDM5C	X	Xp11.22-	X-linked mental retardation	KDM5C

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				p11.21		
115	Test115_SMC1A	SMC1A	X	Xp11.22-p11.21	Cornelia de Lang syndrome 2 (CDLS2)	SMC1A
116	Test116_FTSJ1	FTSJ1	X	Xp11.23	X-linked mental retardation	FTSJ1
117	Test117_PORCN	PORCN	X	Xp11.23	Focal Dermal Hypoplasia	PORCN
118	Test118_PQBP1	PQBP1	X	Xp11.23	X-linked mental retardation	PQBP1
119	Test119_SLC38A5	SLC38A5	X	Xp11.23	X-linked mental retardation	SLC38A5
120	Test120_SYN1	SYN1	X	Xp11.23	X-linked mental retardation	SYN1
121	Test121_ZNF41	ZNF41	X	Xp11.23	X-linked mental retardation	ZNF41
122	Test122_ZNF81	ZNF81	X	Xp11.23	X-linked mental retardation	ZNF81
123	Test123_Xp11.23-p11.22	Xp11.23-p11.22	X	Xp11.23-p11.22	Xp11.23-p11.22 duplication syndrome	
124	Test124_ZNF674	ZNF674	X	Xp11.3	X-linked mental retardation	ZNF674
125	Test125_CASK	CASK	X	Xp11.4	X-linked mental retardation and FG syndrome	CASK
126	Test126_OTC	OTC	X	Xp11.4	Ornithine transcarbamylase (OTC) deficiency	OTC
127	Test127_TSPAN7	TSPAN7	X	Xp11.4	X-linked mental retardation	TSPAN7
128	Test128_ARX	ARX	X	Xp21	X-linked mental retardation	ARX
129	Test129_CDKL5	CDKL5	X	Xp22	Rett syndrome	CDKL5
130	Test130_RPS6KA3	RPS6KA3	X	Xp22	Coffin-Lowry syndrome (CLS)	RPS6KA3
131	Test131_VCX3A	VCX3A	X	Xp22	X-linked mental retardation	VCX3A
132	Test132_SMS	SMS	X	Xp22.1	Snyder-Robinson syndrome	SMS
133	Test133_PTCHD1	PTCHD1	X	Xp22.11	Xp22 Deletion syndrome	PTCHD1
134	Test134_IL1RAPL1	IL1RAPL1	X	Xp22.1-p21.3	X-linked mental retardation	IL1RAPL1
135	Test135_AP1S2	AP1S2	X	Xp22.2	X-linked mental retardation	AP1S2
136	Test136_FANCB	FANCB	X	Xp22.2	VACTERL-H syndrome	FANCB
137	Test137_OFD1	OFD1	X	Xp22.2	Joubert syndrome	OFD1
138	Test138_NLGN4	NLGN4	X	Xp22.32	Autism Spectrum Disorders	NLGN4
139	Test139_TM4SF2	TM4SF2	X	Xq11	X-linked mental retardation	TM4SF2
140	Test140_ARHGEF9	ARHGEF9	X	Xq11.1	X-linked mental retardation	ARHGEF9
141	Test141_ATP6AP2	ATP6AP2	X	Xq12	X-linked mental retardation	ATP6AP2
142	Test142_OPHN1	OPHN1	X	Xq12	X-linked mental retardation	OPHN1
143	Test143_DLG3	DLG3	X	Xq13.1	X-linked mental retardation	DLG3

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144	Test144_NLGN3	NLGN3	X	Xq13.1	Autism Spectrum Disorders	NLGN3
145	Test145_SLC16A2	SLC16A2	X	Xq13.2	X-linked mental retardation	SLC16A2
146	Test146_KIAA2022	KIAA2022	X	Xq13.3	X-linked mental retardation	KIAA2022
147	Test147_ATRX	ATRX	X	Xq21.1	X-linked mental retardation	ATRX
148	Test148_BRWD3	BRWD3	X	Xq21.1	X-linked mental retardation	BRWD3
149	Test149_SRPX2	SRPX2	X	Xq21.33-q23	X-linked mental retardation	SRPX2
150	Test150_PLP1	PLP1	X	Xq22	Pelizaeus-Merzbacher disease (PMD)	PLP1
151	Test151_TIMM8A	TIMM8A	X	Xq22.1	X-linked mental retardation	TIMM8A
152	Test152_DCX	DCX	X	Xq22.3	X-linked lissencephaly-1 (LISX)	DCX
153	Test153_ACSL4	ACSL4	X	Xq22.3-q23	X-linked mental retardation	ACSL4
154	Test154_AGTR2	AGTR2	X	Xq22-q23	X-linked mental retardation	AGTR2
155	Test155_CUL4B	CUL4B	X	Xq23	X-linked mental retardation	CUL4B
156	Test156_PAK3	PAK3	X	Xq23	X-linked mental retardation	PAK3
157	Test157_UBE2A	UBE2A	X	Xq24	X-linked mental retardation	UBE2A
158	Test158_ZCCHC12	ZCCHC12	X	Xq24	X-linked mental retardation	ZCCHC12
159	Test159_GRIA3	GRIA3	X	Xq25	X-linked mental retardation	GRIA3
160	Test160_UPF3B	UPF3B	X	Xq25-q26	X-linked mental retardation	UPF3B
161	Test161_OCRL1	OCRL1	X	Xq25-q26.1	Lowe syndrome	OCRL1
162	Test162_ZDHHC9	ZDHHC9	X	Xq26.1	X-linked mental retardation	ZDHHC9
163	Test163_HPRT1	HPRT1	X	Xq26.2	Lesch-Nyhan syndrome	HPRT1
164	Test164_ARHGEF6	ARHGEF6	X	Xq26.3	X-linked mental retardation	ARHGEF6
165	Test165_PHF6	PHF6	X	Xq26.3	X-linked mental retardation	PHF6
166	Test166_SLC9A6	SLC9A6	X	Xq26.3	X-linked mental retardation	SLC9A6
167	Test167_SOX3	SOX3	X	Xq27.1	X-linked mental retardation	SOX3
168	Test168_FMR1	FMR1	X	Xq27.3	Fragile X	FMR1
169	Test169_ABCD1	ABCD1	X	Xq28	Xq28 Deletion syndrome	ABCD1
170	Test170_AFF2	AFF2	X	Xq28	X-linked mental retardation	AFF2
171	Test171_GDI1	GDI1	X	Xq28	X-linked mental retardation	GDI1
172	Test172_MECP2	MECP2	X	Xq28	Rett syndrome	MECP2
173	Test173_RAB39B	RAB39B	X	Xq28	X-linked mental retardation	RAB39B
174	Test174_RPL10	RPL10	X	Xq28	X-linked mental retardation	RPL10
175	Test175_SLC6A8	SLC6A8	X	Xq28	X-linked mental retardation	SLC6A8

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176	Test176_Karyotype	Karyotype	all			
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Website link to updated table: http://ndar.nih.gov/ndarpublicweb/Documents/Genetic_Tests.pdf