



ChiLDReNLink: LOGIC

Form 04B Genetics Testing Results LOGIC

B: GENE ANALYSIS

B1	DNA Test Date:	____ / ____ / ____		
B2	Gene(s) Analyzed			
B2a	ABCB11	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2b	ABCB4	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2c	ABCC2	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2d	AKR1D1	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2e	AMACR	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2f	ATP8B1	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2g	BAAT	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2h	BCS1L	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2i	CLDN1	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2k	CYP7A1	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2l	CYP7B1	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2m	DCDC2	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2n	GNAS	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2o	HSD3B7	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2p	JAG1	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2q	MYO5B	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2r	NOTCH2	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2s	NPC1	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done

B: GENE ANALYSIS

B2t	NPC2	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2u	NR1H4	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2v	SLC25A13	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2w	SLC27A5	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2x	TALDO1	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2y	TJP2	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2z	UGT1A1	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2aa	VIPAS39	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B2ab	VPS33B	<input type="radio"/> No	<input type="radio"/> Yes	<input type="radio"/> Not Done
B3	Methods used (check all that apply):	<input type="checkbox"/> MLPA <input type="checkbox"/> SNP array <input type="checkbox"/> Targeted NGS <input type="checkbox"/> QPCR <input type="checkbox"/> Capillary Sequencing, all coding exons <input type="checkbox"/> Capillary Sequencing, partial <input type="checkbox"/> Other, specify: _____		

Add Mutation to provide information for each gene tested. If multiple tests were conducted for the same gene, add a new row for each test.

B4: Gene Analyzed	<input type="checkbox"/> ABCB11	<input type="checkbox"/> ABCB4	<input type="checkbox"/> ABCB11	<input type="checkbox"/> ABCB4	<input type="checkbox"/> ABCB11	<input type="checkbox"/> ABCB4	<input type="checkbox"/> ABCB11	<input type="checkbox"/> ABCB4	<input type="checkbox"/> ABCB11	<input type="checkbox"/> ABCB4
	<input type="checkbox"/> ABCC2	<input type="checkbox"/> AKR1D1	<input type="checkbox"/> ABCC2	<input type="checkbox"/> AKR1D1	<input type="checkbox"/> ABCC2	<input type="checkbox"/> AKR1D1	<input type="checkbox"/> ABCC2	<input type="checkbox"/> AKR1D1	<input type="checkbox"/> ABCC2	<input type="checkbox"/> AKR1D1
	<input type="checkbox"/> AMACR	<input type="checkbox"/> ATP8B1	<input type="checkbox"/> AMACR	<input type="checkbox"/> ATP8B1	<input type="checkbox"/> AMACR	<input type="checkbox"/> ATP8B1	<input type="checkbox"/> AMACR	<input type="checkbox"/> ATP8B1	<input type="checkbox"/> AMACR	<input type="checkbox"/> ATP8B1
	<input type="checkbox"/> BAAT	<input type="checkbox"/> BCS1L	<input type="checkbox"/> BAAT	<input type="checkbox"/> BCS1L	<input type="checkbox"/> BAAT	<input type="checkbox"/> BCS1L	<input type="checkbox"/> BAAT	<input type="checkbox"/> BCS1L	<input type="checkbox"/> BAAT	<input type="checkbox"/> BCS1L
	<input type="checkbox"/> CLDN1	<input type="checkbox"/> CYP27A1	<input type="checkbox"/> CLDN1	<input type="checkbox"/> CYP27A1	<input type="checkbox"/> CLDN1	<input type="checkbox"/> CYP27A1	<input type="checkbox"/> CLDN1	<input type="checkbox"/> CYP27A1	<input type="checkbox"/> CLDN1	<input type="checkbox"/> CYP27A1
	<input type="checkbox"/> CYP7A1	<input type="checkbox"/> CYP7B1	<input type="checkbox"/> CYP7A1	<input type="checkbox"/> CYP7B1	<input type="checkbox"/> CYP7A1	<input type="checkbox"/> CYP7B1	<input type="checkbox"/> CYP7A1	<input type="checkbox"/> CYP7B1	<input type="checkbox"/> CYP7A1	<input type="checkbox"/> CYP7B1
	<input type="checkbox"/> DCDC2	<input type="checkbox"/> GNAS	<input type="checkbox"/> DCDC2	<input type="checkbox"/> GNAS	<input type="checkbox"/> DCDC2	<input type="checkbox"/> GNAS	<input type="checkbox"/> DCDC2	<input type="checkbox"/> GNAS	<input type="checkbox"/> DCDC2	<input type="checkbox"/> GNAS
	<input type="checkbox"/> HSD3B7	<input type="checkbox"/> JAG1	<input type="checkbox"/> HSD3B7	<input type="checkbox"/> JAG1	<input type="checkbox"/> HSD3B7	<input type="checkbox"/> JAG1	<input type="checkbox"/> HSD3B7	<input type="checkbox"/> JAG1	<input type="checkbox"/> HSD3B7	<input type="checkbox"/> JAG1
	<input type="checkbox"/> MYO5B	<input type="checkbox"/> NOTCH2	<input type="checkbox"/> MYO5B	<input type="checkbox"/> NOTCH2	<input type="checkbox"/> MYO5B	<input type="checkbox"/> NOTCH2	<input type="checkbox"/> MYO5B	<input type="checkbox"/> NOTCH2	<input type="checkbox"/> MYO5B	<input type="checkbox"/> NOTCH2
	<input type="checkbox"/> NPC1	<input type="checkbox"/> NPC2	<input type="checkbox"/> NPC1	<input type="checkbox"/> NPC2	<input type="checkbox"/> NPC1	<input type="checkbox"/> NPC2	<input type="checkbox"/> NPC1	<input type="checkbox"/> NPC2	<input type="checkbox"/> NPC1	<input type="checkbox"/> NPC2
	<input type="checkbox"/> NR1H4	<input type="checkbox"/> SLC25A13	<input type="checkbox"/> NR1H4	<input type="checkbox"/> SLC25A13	<input type="checkbox"/> NR1H4	<input type="checkbox"/> SLC25A13	<input type="checkbox"/> NR1H4	<input type="checkbox"/> SLC25A13	<input type="checkbox"/> NR1H4	<input type="checkbox"/> SLC25A13
	<input type="checkbox"/> SLC27A5	<input type="checkbox"/> TALDO1	<input type="checkbox"/> SLC27A5	<input type="checkbox"/> TALDO1	<input type="checkbox"/> SLC27A5	<input type="checkbox"/> TALDO1	<input type="checkbox"/> SLC27A5	<input type="checkbox"/> TALDO1	<input type="checkbox"/> SLC27A5	<input type="checkbox"/> TALDO1
	<input type="checkbox"/> TJP2	<input type="checkbox"/> UGT1A1	<input type="checkbox"/> TJP2	<input type="checkbox"/> UGT1A1	<input type="checkbox"/> TJP2	<input type="checkbox"/> UGT1A1	<input type="checkbox"/> TJP2	<input type="checkbox"/> UGT1A1	<input type="checkbox"/> TJP2	<input type="checkbox"/> UGT1A1
	<input type="checkbox"/> VIPAS39	<input type="checkbox"/> VPS33B	<input type="checkbox"/> VIPAS39	<input type="checkbox"/> VPS33B	<input type="checkbox"/> VIPAS39	<input type="checkbox"/> VPS33B	<input type="checkbox"/> VIPAS39	<input type="checkbox"/> VPS33B	<input type="checkbox"/> VIPAS39	<input type="checkbox"/> VPS33B
B5: Mutation Detected (check all that apply):	<input type="checkbox"/> cDNA change, specify: _____	<input type="checkbox"/> cDNA change, specify: _____	<input type="checkbox"/> cDNA change, specify: _____	<input type="checkbox"/> cDNA change, specify: _____	<input type="checkbox"/> cDNA change, specify: _____					
	<input type="checkbox"/> Protein change, specify: _____	<input type="checkbox"/> Protein change, specify: _____	<input type="checkbox"/> Protein change, specify: _____	<input type="checkbox"/> Protein change, specify: _____	<input type="checkbox"/> Protein change, specify: _____					
B6: Change Type:	<input type="radio"/> Homozygous <input type="radio"/> Heterozygous	<input type="radio"/> Homozygous <input type="radio"/> Heterozygous	<input type="radio"/> Homozygous <input type="radio"/> Heterozygous	<input type="radio"/> Homozygous <input type="radio"/> Heterozygous	<input type="radio"/> Homozygous <input type="radio"/> Heterozygous					

B: GENE ANALYSIS					
B7: Disease Causing:	<input type="radio"/> Definite <input type="radio"/> Possible <input type="radio"/> Unlikely	<input type="radio"/> Definite <input type="radio"/> Possible <input type="radio"/> Unlikely	<input type="radio"/> Definite <input type="radio"/> Possible <input type="radio"/> Unlikely	<input type="radio"/> Definite <input type="radio"/> Possible <input type="radio"/> Unlikely	<input type="radio"/> Definite <input type="radio"/> Possible <input type="radio"/> Unlikely
B8: Classification:	<input type="radio"/> Duplication <input type="radio"/> Insertion/Deletion <input type="radio"/> Missense <input type="radio"/> Nonsense <input type="radio"/> Splice site <input type="radio"/> Synonymous <input type="radio"/> Whole/partial gene deletion <input type="radio"/> Other (specify): _____	<input type="radio"/> Duplication <input type="radio"/> Insertion/Deletion <input type="radio"/> Missense <input type="radio"/> Nonsense <input type="radio"/> Splice site <input type="radio"/> Synonymous <input type="radio"/> Whole/partial gene deletion <input type="radio"/> Other (specify): _____	<input type="radio"/> Duplication <input type="radio"/> Insertion/Deletion <input type="radio"/> Missense <input type="radio"/> Nonsense <input type="radio"/> Splice site <input type="radio"/> Synonymous <input type="radio"/> Whole/partial gene deletion <input type="radio"/> Other (specify): _____	<input type="radio"/> Duplication <input type="radio"/> Insertion/Deletion <input type="radio"/> Missense <input type="radio"/> Nonsense <input type="radio"/> Splice site <input type="radio"/> Synonymous <input type="radio"/> Whole/partial gene deletion <input type="radio"/> Other (specify): _____	<input type="radio"/> Duplication <input type="radio"/> Insertion/Deletion <input type="radio"/> Missense <input type="radio"/> Nonsense <input type="radio"/> Splice site <input type="radio"/> Synonymous <input type="radio"/> Whole/partial gene deletion <input type="radio"/> Other (specify): _____
B9: Father sample available?	<input type="radio"/> No <input type="radio"/> Yes (specify K#): _____	<input type="radio"/> No <input type="radio"/> Yes (specify K#): _____	<input type="radio"/> No <input type="radio"/> Yes (specify K#): _____	<input type="radio"/> No <input type="radio"/> Yes (specify K#): _____	<input type="radio"/> No <input type="radio"/> Yes (specify K#): _____
B10: Mutation detected in father?	<input type="radio"/> No, mutation not detected in father <input type="radio"/> Yes <input type="radio"/> Not Done	<input type="radio"/> No, mutation not detected in father <input type="radio"/> Yes <input type="radio"/> Not Done	<input type="radio"/> No, mutation not detected in father <input type="radio"/> Yes <input type="radio"/> Not Done	<input type="radio"/> No, mutation not detected in father <input type="radio"/> Yes <input type="radio"/> Not Done	<input type="radio"/> No, mutation not detected in father <input type="radio"/> Yes <input type="radio"/> Not Done
B11: Mother sample available?	<input type="radio"/> No <input type="radio"/> Yes (specify K#): _____	<input type="radio"/> No <input type="radio"/> Yes (specify K#): _____	<input type="radio"/> No <input type="radio"/> Yes (specify K#): _____	<input type="radio"/> No <input type="radio"/> Yes (specify K#): _____	<input type="radio"/> No <input type="radio"/> Yes (specify K#): _____
B12: Mutation detected in mother?	<input type="radio"/> No, mutation not detected in mother <input type="radio"/> Yes <input type="radio"/> Not Done	<input type="radio"/> No, mutation not detected in mother <input type="radio"/> Yes <input type="radio"/> Not Done	<input type="radio"/> No, mutation not detected in mother <input type="radio"/> Yes <input type="radio"/> Not Done	<input type="radio"/> No, mutation not detected in mother <input type="radio"/> Yes <input type="radio"/> Not Done	<input type="radio"/> No, mutation not detected in mother <input type="radio"/> Yes <input type="radio"/> Not Done
B13: Mutation Inheritance (check all that apply):	<input type="checkbox"/> Paternal <input type="checkbox"/> Maternal <input type="checkbox"/> de novo <input type="checkbox"/> Unknown _____	<input type="checkbox"/> Paternal <input type="checkbox"/> Maternal <input type="checkbox"/> de novo <input type="checkbox"/> Unknown _____	<input type="checkbox"/> Paternal <input type="checkbox"/> Maternal <input type="checkbox"/> de novo <input type="checkbox"/> Unknown _____	<input type="checkbox"/> Paternal <input type="checkbox"/> Maternal <input type="checkbox"/> de novo <input type="checkbox"/> Unknown _____	<input type="checkbox"/> Paternal <input type="checkbox"/> Maternal <input type="checkbox"/> de novo <input type="checkbox"/> Unknown _____
B14: Classification (check all that apply):	Published > 2 or known <input type="checkbox"/> Severe <input type="checkbox"/> Mild <input type="checkbox"/> Studied in vitro <input type="checkbox"/> Possible/probable SNP	Published > 2 or known <input type="checkbox"/> Severe <input type="checkbox"/> Mild <input type="checkbox"/> Studied in vitro <input type="checkbox"/> Possible/probable SNP	Published > 2 or known <input type="checkbox"/> Severe <input type="checkbox"/> Mild <input type="checkbox"/> Studied in vitro <input type="checkbox"/> Possible/probable SNP	Published > 2 or known <input type="checkbox"/> Severe <input type="checkbox"/> Mild <input type="checkbox"/> Studied in vitro <input type="checkbox"/> Possible/probable SNP	Published > 2 or known <input type="checkbox"/> Severe <input type="checkbox"/> Mild <input type="checkbox"/> Studied in vitro <input type="checkbox"/> Possible/probable SNP
B15: Comments:					

G: INVESTIGATOR SIGNATURE

G1	Investigator Signed?	<input type="radio"/> No → Done <input type="radio"/> Yes _____
G2	Date investigator signed	____ / ____ / ____