Patient:		DOB:	8/21/2019	Lab #:	19040699PN

Table of Residual Risks by Ethnicity

Please note: This table displays the residual risk of being affected with the disease after a negative result. **If a patient is reported to be affected with a disease, this table does not apply for that disease.**

Disease (Inheritance)	Gene	Ethnicity	Disease Frequency	Genotype Detection Rate	Residual Risk of Being Affected	Analytical Genotype Detection Rate
Abetalipoproteinemia (AR)	*MTTP*	African	<1 in 1,000,000	94%	1 in 16,900,000	94%
NM_000253.3		Ashkenazi Jewish	1 in 120,000	94%	1 in 2,100,000	
		East Asian	<1 in 1,000,000	65%	1 in 2,900,000	
		Caucasian	<1 in 1,000,000	63%	1 in 2,700,000	
		Latino	<1 in 1,000,000	94%	1 in 16,900,000	
		South Asian	<1 in 1,000,000	94%	1 in 16,900,000	
		Worldwide	<1 in 1,000,000	73%	1 in 3,700,000	
Acrodermatitis Enteropathica (AR)	*SLC39A4*	African	1 in 710,000	96%	1 in 17,900,000	96%
NM_130849.3		East Asian	<1 in 1,000,000	96%	1 in 25,300,000	
		Finnish	1 in 190,000	96%	1 in 4,700,000	
		Caucasian	1 in 400,000	96%	1 in 7,800,000	
		Latino	1 in 1,000,000	82%	1 in 5,500,000	
		South Asian	<1 in 1,000,000	96%	1 in 25,300,000	
		Worldwide	1 in 600,000	93%	1 in 9,000,000	
Acute Infantile Liver Failure (AR)	*TRMU*	African	<1 in 1,000,000	79%	1 in 4,700,000	98%
NM_018006.4		Ashkenazi Jewish	1 in 840,000	98%	1 in 42,400,000	
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	84%	1 in 6,200,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	1 in 410,000	61%	1 in 1,100,000	
		Worldwide	<1 in 1,000,000	79%	1 in 4,800,000	
		Sephardic Jewish, Yemenite	1 in 4,600	66%	1 in 13,500	
Adenosine Deaminase Deficiency (AR)	*ADA*	African	1 in 33,000	85%	1 in 230,000	98%
NM_000022.2		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	1 in 610,000	85%	1 in 4,200,000	
		Latino	1 in 250,000	91%	1 in 2,900,000	
		South Asian	1 in 320,000	75%	1 in 1,300,000	
		Worldwide	1 in 370,000	83%	1 in 2,100,000	
Adrenoleukodystrophy, X-Linked (XL)	*ABCD1*	Worldwide	1 in 20,000	47%	1 in 38,000	89%
NM_000033.3						
Exception: Exons 8 and 9						
Alagille Syndrome 1 / Tetralogy of Fallot (AD)	*JAG1*	Worldwide	1 in 30,000	80%	1 in 150,000	99%
NM_000214.2						
Alpha-Mannosidosis (AR)	*MAN2B1*	African	1 in 340,000	98%	1 in 17,000,000	98%
NM_000528.3		East Asian	<1 in 1,000,000	77%	1 in 4,300,000	
		Finnish	1 in 190,000	98%	1 in 9,600,000	
		Caucasian	1 in 770,000	86%	1 in 5,700,000	
		Latino	<1 in 1,000,000	76%	1 in 4,200,000	
		South Asian	<1 in 1,000,000	47%	1 in 1,900,000	
		Worldwide	1 in 720,000	86%	1 in 5,300,000	
Alpha-Thalassemia (AR)	*HBA1 / HBA2*	Caucasian	<1 in 1,000,000	98%	1 in 20,000,000	98%
NM_000558.4 / NM_000517.4		Southeast Asian	1 in 50	98%	1 in 1,000	
		East Asian	1 in 650	98%	1 in 13,000	
		Worldwide	1 in 8,800	98%	1 in 180,000	

Patient:		DOB:	8/21/2019	Lab #:	19040699PN

Alport Syndrome (*COL4A3*-Related) (AR)	*COL4A3*	African	1 in 430,000	72%	1 in 1,600,000	98%
NM_000091.4		Ashkenazi Jewish	1 in 210,000	98%	1 in 10,300,000	
		East Asian	1 in 230,000	74%	1 in 900,000	
		Finnish	<1 in 1,000,000	65%	1 in 2,900,000	
		Caucasian	1 in 190,000	77%	1 in 830,000	
		Latino	1 in 150,000	77%	1 in 660,000	
		South Asian	1 in 520,000	80%	1 in 2,700,000	
		Worldwide	1 in 220,000	79%	1 in 1,000,000	
Alport Syndrome (*COL4A4*-Related) (AR)	*COL4A4*	African	1 in 540,000	57%	1 in 1,300,000	96%
NM_000092.4		Ashkenazi Jewish	<1 in 1,000,000	96%	1 in 25,300,000	
		East Asian	1 in 100,000	48%	1 in 190,000	
		Finnish	<1 in 1,000,000	96%	1 in 25,300,000	
		Caucasian	1 in 490,000	66%	1 in 1,400,000	
		Latino	1 in 520,000	88%	1 in 4,300,000	
		South Asian	1 in 690,000	86%	1 in 4,900,000	
		Worldwide	1 in 510,000	65%	1 in 1,500,000	
Alport Syndrome (*COL4A5*-Related) (XL)	*COL4A5*	Worldwide	1 in 60,000	80%	1 in 300,000	94%
NM_000495.3						
Argininemia (AR)	*ARG1*	African	<1 in 1,000,000	72%	1 in 3,600,000	90%
NM_000045.3		Ashkenazi Jewish	<1 in 1,000,000	9[illegible]	1 in 10,300,000	
		East Asian	<1 in 1,000,000	[illegible]%	1 in 1,200,000	
		Caucasian	<1 in 1,000,000	41%	1 in 1,700,000	
		Latino	1 in [illegible],000	43%	1 in 960,000	
		South Asian	1 in 1,[illegible]000	40%	1 in 1,700,000	
		Worldwide	<1 in 1,000,000	45%	1 in 1,800,000	
Argininosuccinic Aciduria (AR)	*ASL*	African	1 in 560,000	49%	1 in 1,100,000	98%
NM_000048.3		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		East Asian	1 in 790,000	79%	1 in 3,700,000	
		Finnish	1 in 33,000	98%	1 in 1,700,000	
		Caucasian	1 in 55,000	81%	1 in 290,000	
		[illegible]	1 in 760,000	51%	1 in 1,600,000	
		South Asian	<1 in 1,000,000	67%	1 in 3,100,000	
		Worldwide	1 in 100,000	77%	1 in 450,000	
Ataxia With Isolated Vitamin E Deficiency (AR)	*TTPA*	African	1 in 410,000	98%	1 in 20,500,000	98%
NM_000370.3		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	98%	1 in 50,300,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	1 in 150,000	98%	1 in 7,700,000	
Barth Syndrome (XL)	[illegible]	Worldwide	1 in 150,000	59%	1 in 370,000	97%
NM_000495.3						
Beta-Globin Related Hemoglobinopathies (AD/AR)	*HBB*	African	1 in 40,000	85%	1 in 267,000	98%
NM_000518.4		Ashkenazi Jewish	1 in 570,000	98%	1 in 28,500,000	
		East Asian	1 in 30,000	87%	1 in 231,000	
		Finnish	<1 in 1,000,000	23%	1 in 1,300,000	
		Caucasian	1 in 210,000	80%	1 in 1,050,000	
		Latino	<1 in 1,000,000	79%	1 in 4,760,000	
		South Asian	1 in 3,000	95%	1 in 60,000	
		Worldwide	1 in 30,000	91%	1 in 333,000	
		Mediterranean	1 in 3,100	>90%	1 in 31,000	
Beta-Globin Related Hemoglobinopathies: Sickling Disease (HbS and HbC) (AR)	*HBB*	African	1 in 300	>99%	1 in 30,000	>99%
NM_000518.4		Caucasian	<1 in 1,000,000	>99%	1 in 100,000,000	
		Latino	1 in 180,000	>99%	1 in 18,000,000	
		South Asian	<1 in 1,000,000	>99%	1 in 100,000,000	
Variants Tested: c.19G>A, p.E7K; c.20A>T, p.E7V		Worldwide	1 in 30,000	>99%	1 in 3,300,000	

Patient:		DOB:	8/21/2019	Lab #:	19040699PN

3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency (AR) NM_000198.3	*HSD3B2*	African	<1 in 1,000,000	79%	1 in 4,700,000	98%
		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	55%	1 in 2,200,000	
		Latino	<1 in 1,000,000	48%	1 in 1,900,000	
		South Asian	<1 in 1,000,000	74%	1 in 3,800,000	
		Worldwide	<1 in 1,000,000	63%	1 in 2,700,000	
Beta-Ketothiolase Deficiency (AR) NM_000019.3	*ACAT1*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		East Asian	1 in 340,000	85%	1 in 2,300,000	
		Caucasian	<1 in 1,000,000	78%	1 in 4,600,000	
		Latino	1 in 120,000	92%	1 in 1,500,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	86%	1 in 7,000,000	
Beta-Mannosidosis (AR) NM_005908.3	*MANBA*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	1 in 960,000	90%	1 in 9,100,000	
		Latino	<1 in 1,000,000	90%	1 in 10,200,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	9[illegible]%	1 in 13,200,000	
BH4-Deficient Hyperphenylalaninemia C (AR) NM_000320.2	*QDPR*	Caucasian	<1 in 1,000,000	[illegible]%	1 in 1,400,000	98%
		Latino	<1 in 1,000,000	11%	1 in 1,100,000	
		South Asian	<1 in 1,000,000	44%	1 in 1,800,000	
		Worldwide	<1 in 1,000,000	30%	1 in 1,400,000	
BH4-Deficient Hyperphenylalaninemia D (AR) NM_000281.3	*PCBD1*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		Ashkenazi Jewish	1 in 710,000	98%	1 in 35,900,000	
		Caucasian	<1 in 1,000,000	66%	1 in 3,000,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	76%	1 in 4,200,000	
Biotinidase Deficiency (AR) NM_000060.3	*BTD*	African	1 in 11,000	87%	1 in 86,000	98%
		Ashkenazi Jewish	1 in 950	98%	1 in 47,000	
		East Asian	1 in 420,000	84%	1 in 2,600,000	
		Finnish	1 in 330	98%	1 in 16,000	
		Caucasian	1 in 540	96%	1 in 13,000	
		Latino	1 in 2,400	94%	1 in 38,000	
		South Asian	1 in 210	97%	1 in 6,200	
		Worldwide	1 in 690	96%	1 in 16,000	
Canavan Disease (AR) NM_000049.2	*ASPA*	African	<1 in 1,000,000	96%	1 in 25,300,000	96%
		Ashkenazi Jewish	1 in 9,900	96%	1 in 250,000	
		Finnish	1 in 230,000	96%	1 in 5,900,000	
		Caucasian	1 in 950,000	77%	1 in 4,100,000	
		Latino	<1 in 1,000,000	76%	1 in 4,200,000	
		South Asian	<1 in 1,000,000	38%	1 in 1,600,000	
		Worldwide	1 in 620,000	85%	1 in 4,200,000	
Carbamoylphosphate Synthetase I Deficiency (AR) NM_001875.4	*CPS1*	African	1 in 640,000	39%	1 in 1,100,000	96%
		Ashkenazi Jewish	<1 in 1,000,000	96%	1 in 25,300,000	
		East Asian	1 in 200,000	46%	1 in 370,000	
		Finnish	<1 in 1,000,000	53%	1 in 2,100,000	
		Caucasian	1 in 470,000	47%	1 in 880,000	
		Latino	<1 in 1,000,000	41%	1 in 1,700,000	
		South Asian	<1 in 1,000,000	21%	1 in 1,300,000	
		Worldwide	1 in 690,000	45%	1 in 1,300,000	
Carnitine Acylcarnitine Translocase Deficiency (AR) NM_001875.4	*SLC25A20*	African	<1 in 1,000,000	63%	1 in 2,700,000	85%
		East Asian	<1 in 1,000,000	85%	1 in 6,500,000	
		Caucasian	<1 in 1,000,000	49%	1 in 1,900,000	
		Latino	<1 in 1,000,000	12%	1 in 1,100,000	
		South Asian	<1 in 1,000,000	7%	1 in 1,100,000	
		Worldwide	<1 in 1,000,000	37%	1 in 1,600,000	

Patient:		DOB:	8/21/2019	Lab #:	19040699PN

Carnitine Palmitoyltransferase IA Deficiency (AR) NM_001876.3	*CPT1A*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		Ashkenazi Jewish	1 in 970,000	98%	1 in 48,500,000	
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	1 in 280,000	93%	1 in 4,300,000	
		Caucasian	<1 in 1,000,000	88%	1 in 8,100,000	
		Latino	<1 in 1,000,000	24%	1 in 1,300,000	
		South Asian	<1 in 1,000,000	55%	1 in 2,200,000	
		Worldwide	<1 in 1,000,000	75%	1 in 4,000,000	
		Hutterite	1 in 1,000	>90%	1 in 10,000	
Carnitine Palmitoyltransferase II Deficiency (AR) NM_000098.2	*CPT2*	African	1 in 160,000	72%	1 in 560,000	98%
		Ashkenazi Jewish	1 in 6,700	98%	1 in 340,000	
		East Asian	1 in 280,000	51%	1 in 580,000	
		Finnish	1 in 250,000	98%	1 in 12,400,000	
		Caucasian	1 in 87,000	61%	1 in 220,000	
		Latino	1 in 250,000	87%	1 in 1,900,000	
		South Asian	<1 in 1,000,000	91%	1 in 11,700,000	
		Worldwide	1 in 110,000	73%	1 in 390,000	
Central Hypothyroidism and Testicular Enlargement (XL) NM_001170961.1	*IGSF1*	Worldwide	1 in 500,0	68%	1 in 1,600,000	99%
Cerebral Creatine Deficiency Syndrome 2 (AR) NM_000156.5	*GAMT*	African	<1 in 1,000,000	94%	1 in 2,200,000	96%
		Ashkenazi Jewish	<1 in 1,000,000	96%	1 in 25,300,000	
		East Asian	<1 in 1,000,000	25%	1 in 1,300,000	
		Caucasian	in 760,00	63%	1 in 2,100,000	
		Latino	in 1,000,0	54%	1 in 2,200,000	
		South Asian	<1 in 1,000,000	45%	1 in 1,800,000	
		World	<1 in 1,000,000	50%	1 in 2,000,000	
		Portugu e	1 in 63,000	>90%	1 in 630,000	
Cerebral Creatine Deficiency Syndrome 3 (AR) NM_001482.2	*GATM*	African	1 in 1,000,000	44%	1 in 1,800,000	98%
		st Asian	<1 in 1,000,000	76%	1 in 4,100,000	
		Cauc an	<1 in 1,000,000	16%	1 in 1,200,000	
		W dwide	<1 in 1,000,000	42%	1 in 1,700,000	
Cerebrotendinous Xanthomatosis (AR) NM_000784.3	*CYP27A1*	Afric	1 in 330,000	91%	1 in 3,500,000	98%
		zi Jewish	1 in 440,000	98%	1 in 22,000,000	
		East Asian	1 in 60,000	70%	1 in 200,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	1 in 300,000	86%	1 in 2,200,000	
		Latino	1 in 370,000	85%	1 in 2,400,000	
		South Asian	1 in 82,000	73%	1 in 300,000	
		Worldwide	1 in 210,000	83%	1 in 1,200,000	
		Sephardic Jewish - Moroccan	1 in 23,000	>90%	1 in 230,000	
Chronic Granulomatous Disease (*CYBA*-Related) (AR) NM_000101.2	*CYBA*	African	<1 in 1,000,000	60%	1 in 2,500,000	92%
		Finnish	<1 in 1,000,000	92%	1 in 12,800,000	
		Caucasian	<1 in 1,000,000	43%	1 in 1,800,000	
		Latino	<1 in 1,000,000	92%	1 in 12,800,000	
		South Asian	<1 in 1,000,000	37%	1 in 1,600,000	
		Worldwide	<1 in 1,000,000	49%	1 in 2,000,000	
		Sephardic Jewish - Moroccan	1 in 700	69%	1 in 2,200	
Chronic Granulomatous Disease (*CYBB*-Related) (XL) NM_000397.3	*CYBB*	Worldwide	1 in 100,000	83%	1 in 590,000	98%
Citrin Deficiency (AR) NM_014251.2	*SLC25A13*	African	1 in 760,000	56%	1 in 1,700,000	98%
		Ashkenazi Jewish	1 in 300,000	98%	1 in 15,000,000	
		East Asian	1 in 9,400	96%	1 in 220,000	
		Caucasian	<1 in 1,000,000	90%	1 in 9,800,000	
		Latino	<1 in 1,000,000	87%	1 in 7,600,000	
		South Asian	1 in 990,000	74%	1 in 3,800,000	
		Worldwide	1 in 430,000	87%	1 in 3,200,000	

Patient:		DOB:	8/21/2019	Lab #:	19040699PN

Citrullinemia, Type I (AR)	*ASS1*	African	1 in 460,000	75%	1 in 1,900,000	98%
NM_000050.4		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	1 in 420,000	76%	1 in 1,700,000	
		Latino	1 in 370,000	91%	1 in 4,100,000	
		South Asian	1 in 150,000	73%	1 in 540,000	
		Worldwide	1 in 460,000	76%	1 in 1,900,000	
Combined Pituitary Hormone Deficiency 1 (AD/AR)	*POU1F1*	East Asian	<1 in 1,000,000	3%	1 in 1,000,000	98%
NM_000306.3		Caucasian	<1 in 1,000,000	21%	1 in 1,300,000	
		Latino	<1 in 1,000,000	76%	1 in 4,100,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	76%	1 in 4,100,000	
Combined Pituitary Hormone Deficiency 2 (AR)	*PROP1*	Finnish	<1 in 1,000,000	98%	1 in 50,300,000	98%
NM_006261.4		Caucasian	1 in 930,000	69%	1 in 3,000,000	
		Latino	<1 in 1,000,000	85%	1 in 6,600,000	
		Worldwide	<1 in 1,000,000	74%	1 in 3,800,000	
Combined Pituitary Hormone Deficiency 3 (AR)	*LHX3*	East Asian	<1 in 1,000,000	98%	1 in 50,300,000	98%
NM_014564.3		Caucasian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	98%	1 in 50,300,000	
Congenital Adrenal Hyperplasia due to 11-Beta-Hydroxylase Deficiency (AR)	*CYP11B1*	African	1 in 240,000	29%	1 in 240,000 - 340,000	83%
NM_000497.3		East Asian	<1 in 1,000,000	3%	1 in 1,000,000	
		Caucasian	1 in 720,000	4%	1 in 720,000	
		Latino	1 in 1,300,000	20%	1 in 1,300,000	
		South Asian	1 in 190,000	0-47%	1 in 190,000 - 360,000	
Exception: Exons 3-7		Worldwide	1 in 770,000	2%	1 in 790,000	
Congenital Amegakaryocytic Thrombocytopenia (AR)	*MPL*	African	1 in 990,000	82%	1 in 5,600,000	98%
NM_005373.2		Ashkenazi Jewish	1 in 14,000	98%	1 in 720,000	
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	1 in 230,000	85%	1 in 1,600,000	
		Latino	<1 in 1,000,000	72%	1 in 3,600,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	1 in 360,000	88%	1 in 2,900,000	
Congenital Bile Acid Synthesis Defect (*AKR1D1*-Related) (AR)	*AKR1D1*	East Asian	<1 in 1,000,000	98%	1 in 50,300,000	98%
NM_005989.3		Caucasian	<1 in 1,000,000	77%	1 in 4,400,000	
		South Asian	<1 in 1,000,000	79%	1 in 4,700,000	
		Worldwide	<1 in 1,000,000	80%	1 in 5,100,000	
Congenital Bile Acid Synthesis Defect (*HSD3B7*-Related) (AR)	*HSD3B7*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
(*HSD3B7*-Related) (AR)		Ashkenazi Jewish	<1 in 1,000,000	4%	1 in 1,000,000	
NM_025193.3		East Asian	<1 in 1,000,000	12%	1 in 1,100,000	
		Caucasian	<1 in 1,000,000	71%	1 in 3,400,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	73%	1 in 3,600,000	
		Worldwide	<1 in 1,000,000	72%	1 in 3,500,000	
Congenital Disorder of Glycosylation Type Ib (AR)	*MPI*	African	<1 in 1,000,000	42%	1 in 1,700,000	98%
NM_002435.2		East Asian	1 in 780,000	63%	1 in 2,100,000	
		Finnish	<1 in 1,000,000	66%	1 in 2,900,000	
		Caucasian	1 in 890,000	84%	1 in 5,500,000	
		Latino	<1 in 1,000,000	85%	1 in 6,900,000	
		South Asian	<1 in 1,000,000	55%	1 in 2,200,000	
		Worldwide	<1 in 1,000,000	76%	1 in 4,200,000	
Congenital Hypothyroidism due to Thyroid Dysgenesis or Hypoplasia (AD)	*PAX8*	Worldwide	1 in 30,000	47%	1 in 57,000	99%
NM_003466.3						

Patient:		DOB:	8/21/2019	Lab #:	19040699PN

Congenital Neutropenia (*HAX1*-Related) (AR) NM_006118.3	*HAX1*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	98%	1 in 50,300,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	98%	1 in 50,300,000	
Congenital Nongoitrous Hypothryoidism 1 / Nonautoimmune Hyperthyroidism (AD/AR) NM_000369.2	*TSHR*	African	1 in 670,000	63%	1 in 1,800,000	92%
		Ashkenazi Jewish	1 in 260,000	74%	1 in 990,000	
		East Asian	1 in 34,000	37%	1 in 54,000	
		Finnish	<1 in 1,000,000	92%	1 in 12,800,000	
		Caucasian	1 in 360,000	51%	1 in 720,000	
		Latino	1 in 450,000	57%	1 in 1,000,000	
		South Asian	1 in 300,000	54%	1 in 660,000	
		Worldwide	1 in 280,000	53%	1 in 590,000	
Congenital Nongoitrous Hypothryoidism 4 (AR) NM_000549.4	*TSHB*	Caucasian	<1 in 1,000,000	98%	1 in 50,300,000	98%
		Worldwide	<1 in 1,000,000	98%	1 in 50,300,000	
Congenital Nongoitrous Hypothryoidism 6 (AR) NM_199334.3	*THRA*	Worldwide	1 in 270,000	62%	1 in 710,000	99%
Corticosterone Methyloxidase Deficiency (AR) NM_000498.3	*CYP11B2*	African	<1 in 1,000,000	22%	1 in 1,300,000	67%
		East Asian	<1 in 1,000,000	2%	1 in 1,000,000	
		Finnish	<1 in 1,000,000	3%	1 in 1,000,000	
		Caucasian	<1 in 1,000,000	19%	1 in 1,200,000	
		Latino	<1 in 1,000,000	21%	1 in 1,300,000	
		South Asian	<1 in 1,000,000	17%	1 in 1,200,000	
		Worldwide	<1 in 1,000,000	17%	1 in 1,200,000	
Exception: Exons 3 - 7		Sephardic Jewish, Iranian	1 in 3600	>90%	1 in 36,000	
Crigler-Najjar Syndrome, Types 1 & 2 / Gilbert Syndrome (AR) NM_000463.2	*UGT1A1*	African	<1 in 1,000,000	51%	1 in 2,040,000	94%
		East Asian	1 in 90,000	38%	1 in 145,000	
		Finnish	<1 in 1,000,000	14%	1 in 1,200,000	
		Caucasian	1 in 500,000	33%	1 in 746,000	
		Latino	<1 in 1,000,000	28%	1 in 1,400,000	
		South Asian	1 in 40,000	63%	1 in 108,000	
		Worldwide	1 in 330,000	45%	1 in 600,000	
Cystic Fibrosis (AR) NM_000492.3	*CFTR*	African	1 in 13,000	83%	1 in 78,000	98%
		Ashkenazi Jewish	1 in 2,300	96%	1 in 60,000	
		East Asian	1 in 310,000	65%	1 in 870,000	
		Finnish	1 in 22,000	87%	1 in 180,000	
		Caucasian	1 in 2,100	90%	1 in 22,000	
		Latino	1 in 6,500	92%	1 in 86,000	
		South Asian	1 in 21,000	83%	1 in 120,000	
		Worldwide	1 in 4,500	88%	1 in 37,000	
Cystinosis (AR) NM_004937.2	*CTNS*	African	<1 in 1,000,000	46%	1 in 1,900,000	98%
		East Asian	1 in 620,000	89%	1 in 5,800,000	
		Caucasian	1 in 250,000	94%	1 in 3,900,000	
		Latino	<1 in 1,000,000	79%	1 in 4,800,000	
		South Asian	<1 in 1,000,000	63%	1 in 2,700,000	
		Worldwide	<1 in 1,000,000	82%	1 in 5,600,000	
		French Canadian - Saguenay-Lac St. Jean	1 in 6,000	81%	1 in 32,000	
		Sephardic Jewish - Moroccan	1 in 40,000	85%	1 in 270,000	
Distal Renal Tubular Acidosis and other *SLC4A1*-Related Disorders (AR) NM_000342.3	*SLC4A1*	African	<1 in 1,000,000	62%	1 in 2,600,000	88%
		East Asian	1 in 290,000	49%	1 in 580,000	
		Caucasian	<1 in 1,000,000	27%	1 in 1,400,000	
		Latino	<1 in 1,000,000	88%	1 in 8,600,000	
		South Asian	<1 in 1,000,000	67%	1 in 3,000,000	
		Worldwide	<1 in 1,000,000	41%	1 in 1,700,000	

Patient:		DOB:	8/21/2019	Lab #:	19040699PN

Dopa-Responsive Dystonia / BH4-Deficient Hyperphenylalaninemia B (AD/AR) NM_000161.2	*GCH1*	Worldwide	<1 in 1,000,000	64%	1 in 2,800,000	97%
Dyskeratosis Congenita (*DKC1*-Related) (XL) NM_001363.4	*DKC1*	Worldwide	1 in 5,000,000	73%	1 in 3,700,000	99%
Dyskeratosis Congenita (*RTEL1*-Related) (AR) NM_001283009.1	*RTEL1*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		Ashkenazi Jewish	1 in 49,000	98%	1 in 2,500,000	
		East Asian	1 in 590,000	81%	1 in 3,200,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	84%	1 in 6,400,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	91%	1 in 10,600,000	
Early Infantile Epileptic Encephalopathy 11/ Benign Familial Infantile Seizures 3 (AD) NM_021007.2	*SCN2A*	Worldwide	1 in 50,000	37%	1 in 79,000	98%
Early Infantile Epileptic Encephalopathy 13 / Benign Familial Infantile Seizures 5 (AD) NM_014191.3	*SCN8A*	Worldwide	1 in 7,000	42%	1 in 12,000	99%
Early Infantile Epileptic Encephalopathy 7 / Benign Neonatal Seizures 1 (AD) NM_172107.2	*KCNQ2*	Worldwide	1 in 100,000	61%	1 in 260,000	99%
Ethylmalonic Encephalopathy (AR) NM_014297.3	*ETHE1*	African	<1 in 1,000,000	96%	1 in 25,300,000	96%
		Caucasian	<1 in 1,000,000	39%	1 in 1,600,000	
		Latino	<1 in 1,000,000	86%	1 in 7,000,000	
		South Asian	<1 in 1,000,000	96%	1 in 25,300,000	
		Worldwide	<1 in 1,000,000	59%	1 in 2,400,000	
Fabry Disease (XL) NM_000169.2	*GLA*	Worldwide	1 in 4,000	74%	1 in 15,000	99%
Factor IX Deficiency (XL) NM_000133.3	*F9*	Worldwide	1 in 4,000	61%	1 in 10,000	98%
Familial Hypercholesterolemia (AR) NM_000527.4	*LDLR*	African	1 in 98,000	43%	1 in 170,000	92%
		Ashkenazi Jewish	<1 in 1,000,000	68%	1 in 3,100,000	
		East Asian	1 in 17,000	56%	1 in 39,000	
		Finnish	1 in 340,000	40%	1 in 570,000	
		Caucasian	1 in 56,000	34%	1 in 85,000	
		Latino	1 in 130,000	25%	1 in 180,000	
		South Asian	1 in 69,000	26%	1 in 93,000	
		Worldwide	1 in 64,000	35%	1 in 98,000	
		South African Afrikaner	1 in 20,000	88%	1 in 166,700	
Familial Hyperinsulinemic Hypoglycemia 4 / 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (AR) NM_005327.4	*HADH*	African	<1 in 1,000,000	43%	1 in 1,800,000	92%
		Caucasian	<1 in 1,000,000	65%	1 in 2,900,000	
		Latino	<1 in 1,000,000	23%	1 in 1,300,000	
		Worldwide	<1 in 1,000,000	41%	1 in 1,700,000	
Familial Hyperinsulinism (*ABCC8*-Related) (AR) NM_000352.4	*ABCC8*	African	1 in 260,000	22%	1 in 340,000	98%
		Ashkenazi Jewish	1 in 16,000	78%	1 in 69,000	
		East Asian	1 in 57,000	26%	1 in 76,000	
		Finnish	1 in 180,000	84%	1 in 1,100,000	
		Caucasian	1 in 150,000	33%	1 in 220,000	
		Latino	1 in 320,000	72%	1 in 1,200,000	
		South Asian	1 in 530,000	50%	1 in 1,100,000	
		Worldwide	1 in 140,000	40%	1 in 230,000	

Patient:		DOB:	8/21/2019	Lab #:	19040699PN

Familial Hyperinsulinism (*KCNJ11*-Related) (AR) NM_000525.3	*KCNJ11*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		East Asian	<1 in 1,000,000	6%	1 in 1,100,000	
		Caucasian	<1 in 1,000,000	63%	1 in 2,700,000	
		Latino	<1 in 1,000,000	81%	1 in 5,200,000	
		South Asian	<1 in 1,000,000	75%	1 in 4,000,000	
		Worldwide	<1 in 1,000,000	51%	1 in 2,000,000	
Familial Infantile Convulsions with Paroxysmal Choreoathetosis (AD) NM_145239.2	*PRRT2*	Worldwide	1 in 200,000	77%	1 in 870,000	99%
Fanconi Anemia, Group A (AR) NM_000135.2	*FANCA*	African	1 in 98,000	74%	1 in 370,000	90%
		Ashkenazi Jewish	1 in 250,000	81%	1 in 1,300,000	
		East Asian	1 in 130,000	79%	1 in 650,000	
		Finnish	1 in 290,000	90%	1 in 2,900,000	
		Caucasian	1 in 87,000	76%	1 in 360,000	
		Latino	1 in 310,000	76%	1 in 1,300,000	
		South Asian	1 in 260,000	60%	1 in 660,000	
		Worldwide	1 in 110,000	77%	1 in 470,000	
Fanconi Anemia, Group C (AR) NM_000136.2	*FANCC*	African	1 in 940,0	76%	1 in 3,900,000	98%
		Ashkenazi Jewish	1 in 27,000	98%	1 in 1,300,000	
		East Asian	1 in 470,000	9	1 in 23,700,000	
		Finnish	<1 in 1,000,000	8%	1 in 50,300,000	
		Caucasian	1 740,000	93%	1 in 10,200,000	
		Latino	<1 in 000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,0 000	98%	1 in 50,300,000	
		Worldwide	in 790,0	94%	1 in 12,400,000	
Fanconi Anemia, Group G (AR) NM_001629.1	*FANCG*	African	1 in 980,000	98%	1 in 49,000,000	98%
		East A	1 in 450,000	52%	1 in 940,000	
		Finnis	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasia	in 1,000,000	96%	1 in 25,300,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		ou sian	<1 in 1,000,000	98%	1 in 50,300,000	
		W ldwide	<1 in 1,000,000	91%	1 in 10,500,000	
Fructose-1,6-Bisphosphatase Deficiency (AR) NM_000507.3	*FBP1*	Af n	<1 in 1,000,000	79%	1 in 4,800,000	79%
		East As n	<1 in 1,000,000	79%	1 in 4,800,000	
		Ca sian	<1 in 1,000,000	31%	1 in 1,400,000	
		Latino	<1 in 1,000,000	9%	1 in 1,100,000	
		South Asian	1 in 990,000	52%	1 in 2,000,000	
		Worldwide	<1 in 1,000,000	38%	1 in 1,600,000	
Galactokinase Deficiency (AR) NM_000154.1	*GALK1*	African	1 in 600,000	33%	1 in 900,000	96%
		East Asian	<1 in 1,000,000	30%	1 in 1,400,000	
		Finnish	<1 in 1,000,000	96%	1 in 25,300,000	
		Caucasian	<1 in 1,000,000	52%	1 in 2,100,000	
		Latino	<1 in 1,000,000	61%	1 in 2,600,000	
		South Asian	1 in 640,000	72%	1 in 2,300,000	
		Worldwide	<1 in 1,000,000	55%	1 in 2,200,000	
		Roma	1 in 9,000	>90%	1 in 90,000	
Galactose Epimerase Deficiency (AR) NM_000403.3	*GALE*	African	<1 in 1,000,000	9%	1 in 1,100,000	98%
		East Asian	1 in 43,000	37%	1 in 690,000	
		Caucasian	<1 in 1,000,000	43%	1 in 1,800,000	
		Latino	<1 in 1,000,000	74%	1 in 3,800,000	
		South Asian	<1 in 1,000,000	44%	1 in 1,800,000	
		Worldwide	<1 in 1,000,000	41%	1 in 1,700,000	

Patient:		DOB:	8/21/2019	Lab #:	19040699PN

Galactosemia (AR) NM_000155.3	*GALT*	African	1 in 30,000	87%	1 in 230,000	98%
		Ashkenazi Jewish	1 in 130,000	98%	1 in 8,300,000	
		East Asian	1 in 170,000	22%	1 in 220,000	
		Finnish	<1 in 1,000,000	46%	1 in 1,800,000	
		Caucasian	1 in 60,000	92%	1 in 800,000	
		Latino	1 in 190,000	91%	1 in 2,000,000	
		South Asian	1 in 470,000	81%	1 in 2,500,000	
		Worldwide	1 in 97,000	81%	1 in 500,000	
Gaucher Disease (AR) NM_000157.3	*GBA*	Ashkenazi Jewish	1 in 900	90%	1 in 9,200	90%
		Caucasian	1 in 110,000	76%	1 in 460,000	
		Worldwide	1 in 100,000	75%	1 in 400,000	
Generalized Thyrotropin-Releasing Hormone Resistance (AR) NM_003301.5	*TRHR*	East Asian	<1 in 1,000,000	98%	1 in 50,300,000	98%
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	98%	1 in 50,300,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	98%	1 in 50,300,000	
Glucose Transporter 1 Deficiency Syndrome and Other *SLC2A1*-Related Disorders (AD/AR) NM_006516.2	*SLC2A1*	Worldwide	<1 in 1,000,000	64%	1 in 2,800,000	98%
Glutaric Acidemia, Type I (AR) NM_000159.3	*GCDH*	African	1 in 35,000	70%	1 in 120,000	98%
		East Asian	1 in 40,000	98%	1 in 8,400,000	
		Finnish	1 in 500,000	82%	1 in 2,700,000	
		Caucasian	1 in 160,000	86%	1 in 1,100,000	
		Latino	1 in 290,000	89%	1 in 2,600,000	
		South Asian	1 in 270,000	85%	1 in 1,800,000	
		Worldwide	1 in 160,000	79%	1 in 790,000	
		Oji-Cree First Nations (N. Manitoba)	1 in 300	>90%	1 in 3,000	
		Old Order Amish of Pennsylvania	1 in 500	>90%	1 in 5,000	
		Lumbee Native American	1 in 1,000	>90%	1 in 10,000	
Glutaric Acidemia, Type IIa (AR) NM_000126.3	*ETFA*	African	<1 in 1,000,000	72%	1 in 3,600,000	94%
		East Asian	<1 in 1,000,000	17%	1 in 1,200,000	
		Caucasian	<1 in 1,000,000	67%	1 in 3,000,000	
		Latino	<1 in 1,000,000	60%	1 in 2,500,000	
		South Asian	<1 in 1,000,000	94%	1 in 16,900,000	
		Worldwide	<1 in 1,000,000	69%	1 in 3,300,000	
Glutaric Acidemia, Type IIb (AR) NM_001985.2	*ETFB*	African	<1 in 1,000,000	81%	1 in 5,200,000	98%
		Ashkenazi Jewish	1 in 960,000	98%	1 in 48,500,000	
		Caucasian	<1 in 1,000,000	32%	1 in 1,500,000	
		Latino	1 in 420,000	98%	1 in 20,900,000	
		South Asian	<1 in 1,000,000	42%	1 in 1,700,000	
		Worldwide	<1 in 1,000,000	65%	1 in 2,900,000	
Glutaric Acidemia, Type IIc (AR) NM_004453.3	*ETFDH*	African	1 in 470,000	44%	1 in 840,000	98%
		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		East Asian	1 in 32,000	43%	1 in 56,000	
		Finnish	<1 in 1,000,000	70%	1 in 3,300,000	
		Caucasian	1 in 450,000	64%	1 in 1,300,000	
		Latino	<1 in 1,000,000	34%	1 in 1,500,000	
		South Asian	<1 in 1,000,000	22%	1 in 1,300,000	
		Worldwide	1 in 460,000	51%	1 in 930,000	
Glutathione Synthetase Deficiency (AR) NM_000178.2	*GSS*	African	1 in 490,000	62%	1 in 1,300,000	94%
		Ashkenazi Jewish	1 in 47,000	94%	1 in 790,000	
		East Asian	<1 in 1,000,000	94%	1 in 16,900,000	
		Finnish	<1 in 1,000,000	94%	1 in 16,900,000	
		Caucasian	<1 in 1,000,000	47%	1 in 1,900,000	
		Latino	<1 in 1,000,000	94%	1 in 16,900,000	
		South Asian	<1 in 1,000,000	94%	1 in 16,900,000	
		Worldwide	<1 in 1,000,000	71%	1 in 3,400,000	

Patient:		DOB:	8/21/2019	Lab #:	19040699PN

Glycogen Storage Disease, Type 0 (AR)	*GYS2*	African	1 in 600,000	90%	1 in 6,200,000	90%
NM_021957.3		Ashkenazi Jewish	<1 in 1,000,000	>90%	1 in 10,000,000	
		East Asian	<1 in 1,000,000	90%	1 in 10,300,000	
		Finnish	<1 in 1,000,000	40%	1 in 1,700,000	
		Caucasian	1 in 220,000	64%	1 in 610,000	
		Latino	1 in 130,000	86%	1 in 960,000	
		South Asian	1 in 95,000	90%	1 in 970,000	
		Worldwide	1 in 340,000	71%	1 in 1,200,000	
Glycogen Storage Disease, Type Ia (AR)	*G6PC*	African	<1 in 1,000,000	78%	1 in 4,500,000	98%
NM_000151.3		Ashkenazi Jewish	1 in 22,000	98%	1 in 1,100,000	
		East Asian	1 in 54,000	52%	1 in 110,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	1 in 400,000	88%	1 in 3,500,000	
		Latino	1 in 480,000	79%	1 in 2,300,000	
		South Asian	<1 in 1,000,000	44%	1 in 1,800,000	
		Worldwide	1 in 380,000	82%	1 in 2,100,000	
Glycogen Storage Disease, Type II (AR)	*GAA*	African	1 in 20,000	67%	1 in 60,000	98%
NM_000152.3		Ashkenazi Jewish	1 in 23,00	95%	1 in 460,000	
		East Asian	1 in 16,00	61%	1 in 41,000	
		Finnish	1 in 540,000	35	1 in 820,000	
		Caucasian	1 in 9,600	%	1 in 54,000	
		Latino	in 36,000	74%	1 in 140,000	
		South Asian	1 1,000	83%	1 in 430,000	
		Worldwide	1 in 2 ,000	75%	1 in 81,000	
Glycogen Storage Disease, Type III (AR)	*AGL*	African	in 150,0	73%	1 in 550,000	98%
NM_000028.2		East Asian	1 in 1,000,000	98%	1 in 50,300,000	
		Finni	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucas	1 in 270,000	91%	1 in 3,000,000	
		Latino	1 in 880,000	93%	1 in 12,100,000	
		South Asia	<1 in 1,000,000	53%	1 in 2,100,000	
		W ide	1 in 400,000	84%	1 in 2,500,000	
		Sephardi wish ccan	1 in 5,000	>90%	1 in 50,000	
		Fa se	1 in 3,000	>90%	1 in 30,000	
Glycogen Storage Disease, Type IXb (AR)	*PHKB*	Afric	1 in 790,000	62%	1 in 2,100,000	88%
NM_000293.2		sian	1 in 630,000	88%	1 in 5,400,000	
		Caucasian	1 in 580,000	73%	1 in 2,200,000	
		Latino	1 in 680,000	17%	1 in 820,000	
		South Asian	1 in 580,000	88%	1 in 5,000,000	
		Worldwide	1 in 680,000	66%	1 in 2,000,000	
Glycogen Storage Disease, Type VI (AR)	*PYGL*	African	1 in 510,000	74%	1 in 2,000,000	90%
NM_002863.4		East Asian	1 in 420,000	71%	1 in 1,400,000	
		Finnish	<1 in 1,000,000	51%	1 in 2,000,000	
		Caucasian	1 in 830,000	52%	1 in 1,700,000	
		Latino	<1 in 1,000,000	78%	1 in 4,600,000	
		South Asian	1 in 870,000	19%	1 in 1,100,000	
		Worldwide	1 in 910,000	55%	1 in 2,000,000	
Hemolytic Anemia (*G6PD*-Related) (XL)	*G6PD*	African	1 in 4	91%	1 in 34	99%
NM_001042351.2		Ashkenazi Jewish	1 in 85	95%	1 in 1680	
		East Asian	1 in 18	61%	1 in 45	
		Caucasian	1 in 161	69%	1 in 520	
		Latino	1 in 65	65%	1 in 180	
		South Asian	1 in 11	84%	1 in 64	
		Worldwide	1 in 23	79%	1 in 106	

Patient:		DOB:	8/21/2019	Lab #:	19040699PN

Hereditary Fructose Intolerance (AR)	*ALDOB*	African	1 in 410,000	96%	1 in 10,300,000	96%
NM_000035.3		Ashkenazi Jewish	1 in 79,000	96%	1 in 2,000,000	
		East Asian	<1 in 1,000,000	96%	1 in 25,300,000	
		Finnish	1 in 40,000	96%	1 in 1,000,000	
		Caucasian	1 in 26,000	92%	1 in 320,000	
		Latino	1 in 220,000	88%	1 in 1,900,000	
		South Asian	1 in 620,000	91%	1 in 7,000,000	
		Worldwide	1 in 58,000	92%	1 in 750,000	
HMG-CoA Lyase Deficiency (AR)	*HMGCL*	African	<1 in 1,000,000	96%	1 in 25,300,000	96%
NM_000191.2		East Asian	<1 in 1,000,000	96%	1 in 25,300,000	
		Finnish	<1 in 1,000,000	96%	1 in 25,300,000	
		Caucasian	<1 in 1,000,000	46%	1 in 1,800,000	
		Latino	<1 in 1,000,000	96%	1 in 25,300,000	
		South Asian	<1 in 1,000,000	96%	1 in 25,300,000	
		Worldwide	<1 in 1,000,000	66%	1 in 3,000,000	
HMG-CoA Synthase 2 Deficiency (AR)	*HMGCS2*	African	1 in 520,000	87%	1 in 3,900,000	92%
NM_005518.3		East Asian	1 in 130,000	88%	1 in 1,100,000	
		Caucasian	1 in 910,00	58%	1 in 2,200,000	
		Latino	<1 in 1,000,0	52%	1 in 2,100,000	
		South Asian	1 in 770,000	87	1 in 5,900,000	
		Worldwide	1 in 940,000	%	1 in 2,900,000	
Holocarboxylase Synthetase Deficiency (AR)	*HLCS*	African	<1 in 1,000,000	84%	1 in 6,200,000	98%
NM_000411.6		East Asian	1 in 70,000	90%	1 in 4,900,000	
		Finnish	in 1,0 ,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,	76%	1 in 4,200,000	
		Latino	1 in 1,000,000	75%	1 in 4,000,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	83%	1 in 5,700,000	
Homocystinuria (*CBS*-Related) (AR)	*CBS*	African	1 in 140,000	90%	1 in 1,300,000	94%
NM_000071.2		kenazi Jewish	1 in 440,000	82%	1 in 2,400,000	
		sian	<1 in 1,000,000	53%	1 in 2,100,000	
		innish	1 in 450,000	88%	1 in 3,900,000	
		sian	1 in 80,000	80%	1 in 410,000	
		Latin	1 in 160,000	87%	1 in 1,300,000	
		Asian	<1 in 1,000,000	80%	1 in 5,000,000	
		Worldwide	1 in 130,000	82%	1 in 730,000	
Homocystinuria, Cobalamin E Type (AR)	*MTRR*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
NM_002454.2		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	87%	1 in 7,700,000	
		Latino	1 in 960,000	92%	1 in 12,500,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	91%	1 in 11,200,000	
Homocystinuria-Megaloblastic Anemia, Cobalamin G Type (AR)	*MTR*	African	<1 in 1,000,000	43%	1 in 1,700,000	98%
		Ashkenazi Jewish	<1 in 1,000,000	59%	1 in 2,400,000	
NM_000254.2		East Asian	1 in 490,000	98%	1 in 24,800,000	
		Finnish	<1 in 1,000,000	46%	1 in 1,800,000	
		Caucasian	<1 in 1,000,000	54%	1 in 2,200,000	
		Latino	<1 in 1,000,000	72%	1 in 3,600,000	
		Worldwide	<1 in 1,000,000	57%	1 in 2,300,000	
Hyperinsulinism-Hyperammonemia Syndrome (AD)	*GLUD1*	Worldwide	1 in 6,000	84%	1 in 37,000	99%
NM_005271.3						
Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome (AR)	*SLC25A15*	East Asian	1 in 370,000	98%	1 in 18,400,000	98%
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
NM_014252.3		Caucasian	<1 in 1,000,000	60%	1 in 2,500,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	55%	1 in 2,200,000	
		Worldwide	<1 in 1,000,000	75%	1 in 4,000,000	
		Metis - Saskatchewan	1 in 1,400	>90%	1 in 14,000	

Patient:		DOB:	8/21/2019	Lab #:	19040699PN

Hypophosphatasia (AD/AR)	*ALPL*	African	<1 in 1,000,000	75%	1 in 4,000,000	98%
NM_000478.4		Ashkenazi Jewish	<1 in 1,000,000	44%	1 in 1,800,000	
		East Asian	1 in 69,000	95%	1 in 1,400,000	
		Finnish	1 in 3,100	92%	1 in 39,000	
		Caucasian	1 in 57,000	72%	1 in 200,000	
		Latino	1 in 800,000	24%	1 in 1,100,000	
		South Asian	<1 in 1,000,000	46%	1 in 1,800,000	
		Worldwide	1 in 55,000	79%	1 in 260,000	
		Mennonite	1 in 2,500	>90%	1 in 25,000	
Immunodeficiency 18 (AR)	*CD3E*	Caucasian	<1 in 1,000,000	98%	1 in 50,300,000	98%
NM_000733.3		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	98%	1 in 50,300,000	
Immunodeficiency 19 (AR)	*CD3D*	East Asian	<1 in 1,000,000	98%	1 in 50,300,000	98%
NM_000732.4		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	89%	1 in 9,100,000	
		Latino	<1 in 1,000,000	62%	1 in 2,700,000	
		Worldwide	<1 in 1,000,000	91%	1 in 10,700,000	
Isovaleric Acidemia (AR)	*IVD*	African	1 in 370,0	77%	1 in 1,600,000	98%
NM_002225.3		East Asian	<1 in 1,000,0	61%	1 in 2,600,000	
		Finnish	<1 in 1,000,000	66	1 in 3,000,000	
		Caucasian	1 in 250,000	%	1 in 1,100,000	
		Latino	<1 in 1,000,000	80%	1 in 5,100,000	
		South Asian	<1 in ,000,000	57%	1 in 2,300,000	
		Worldwide	in 40 ,000	77%	1 in 2,000,000	
Krabbe Disease (AR)	*GALC*	African	in 100,0	28%	1 in 140,000	98%
NM_000153.3		Ashkenazi Jewis	1 in 1,000,000	70%	1 in 3,400,000	
		East A n	1 in 6,500	78%	1 in 30,000	
		Finni	1 in 86,000	98%	1 in 4,300,000	
		Caucasi	1 in 19,000	85%	1 in 120,000	
		Latino	1 in 160,000	85%	1 in 1,000,000	
		Sou Asian	1 in 5,000	83%	1 in 30,000	
		rldwi	1 in 24,000	79%	1 in 110,000	
Lipoamide Dehydrogenase Deficiency (AR)	*DLD*	Ashken Jewish	1 in 15,000	98%	1 in 730,000	98%
NM_000108.4		East A n	<1 in 1,000,000	98%	1 in 50,300,000	
		ish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	79%	1 in 4,800,000	
		Latino	<1 in 1,000,000	24%	1 in 1,300,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	87%	1 in 7,800,000	
Lipoid Adrenal Hyperplasia (AR)	*STAR*	African	<1 in 1,000,000	83%	1 in 5,900,000	98%
NM_000349.2		East Asian	1 in 530,000	98%	1 in 26,600,000	
		Finnish	<1 in 1,000,000	50%	1 in 2,000,000	
		Caucasian	<1 in 1,000,000	46%	1 in 1,900,000	
		Latino	<1 in 1,000,000	48%	1 in 1,900,000	
		South Asian	<1 in 1,000,000	66%	1 in 2,900,000	
		Worldwide	<1 in 1,000,000	62%	1 in 2,600,000	
Lipoprotein Lipase Deficiency (AR)	*LPL*	African	1 in 380,000	59%	1 in 930,000	98%
NM_000237.2		East Asian	1 in 43,000	76%	1 in 180,000	
		Caucasian	1 in 560,000	71%	1 in 1,900,000	
		Latino	1 in 560,000	42%	1 in 950,000	
		South Asian	1 in 820,000	25%	1 in 1,100,000	
		Worldwide	1 in 470,000	61%	1 in 1,200,000	
		French Canadian - Saguenay - Lac St. Jean	1 in 8,400	>90%	1 in 84,000	
		French Canadian - Other	1 in 77,000	>90%	1 in 770,000	

Patient:		DOB:	8/21/2019	Lab #:	19040699PN

Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (AR) NM_000182.4	*HADHA*	African	1 in 930,000	61%	1 in 2,400,000	98%
		East Asian	<1 in 1,000,000	61%	1 in 2,500,000	
		Finnish	1 in 60,000	98%	1 in 3,000,000	
		Caucasian	1 in 190,000	93%	1 in 2,600,000	
		Latino	1 in 660,000	89%	1 in 5,900,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	1 in 280,000	90%	1 in 2,700,000	
Lysinuric Protein Intolerance (AR) NM_001126106.2	*SLC7A7*	African	<1 in 1,000,000	66%	1 in 3,000,000	98%
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	1 in 45,000	98%	1 in 2,300,000	
		Caucasian	<1 in 1,000,000	69%	1 in 3,200,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	82%	1 in 5,700,000	
		Worldwide	1 in 810,000	83%	1 in 4,800,000	
		Japanese	1 in 57,000	77%	1 in 250,000	
Malonyl-CoA Decarboxylase Deficiency (AR) NM_012213.2	*MLYCD*	African	<1 in 1,000,000	41%	1 in 1,700,000	74%
		Caucasian	<1 in 1,000,000	44%	1 in 1,800,000	
		Latino	<1 in 1,000,000	29%	1 in 1,400,000	
		South Asian	<1 in 1,000,000	74%	1 in 3,800,000	
		Worldwide	<1 in 1,000,000	51%	1 in 2,100,000	
Maple Syrup Urine Disease, Type 1a (AR) NM_000709.3	*BCKDHA*	African	1 in 910,000	9%	1 in 1,800,000	96%
		Ashkenazi Jewish	1 in 460,000	96%	1 in 11,600,000	
		East Asian	<1 in 1,000,000	61%	1 in 2,600,000	
		Finnish	1 in 1,000,000	96%	1 in 25,300,000	
		Caucasian	<1 in 1,000,000	80%	1 in 4,900,000	
		Latino	1 in 1,000,000	87%	1 in 7,500,000	
		South Asian	<1 in 1,000,000	96%	1 in 25,300,000	
		Worldwide	<1 in 1,000,000	80%	1 in 5,100,000	
		Mennonite	1 in 400	>90%	1 in 4,000	
		Portuguese Roma	1 in 20,000	>90%	1 in 200,000	
Maple Syrup Urine Disease, Type 1b (AR) NM_000056.3	*BCKDHB*	African	<1 in 1,000,000	58%	1 in 2,400,000	98%
		Ashkenazi Jewish	1 in 27,000	98%	1 in 1,300,000	
		East Asian	<1 in 1,000,000	70%	1 in 3,300,000	
		Finnish	1 in 130,000	98%	1 in 6,400,000	
		Caucasian	1 in 370,000	54%	1 in 800,000	
		Latino	1 in 680,000	89%	1 in 6,000,000	
		South Asian	<1 in 1,000,000	60%	1 in 2,500,000	
		Worldwide	1 in 360,000	72%	1 in 1,300,000	
Maple Syrup Urine Disease, Type 2 (AR) NM_001918.3	*DBT*	African	1 in 490,000	92%	1 in 6,300,000	92%
		Ashkenazi Jewish	<1 in 1,000,000	92%	1 in 12,800,000	
		East Asian	1 in 980,000	14%	1 in 1,100,000	
		Finnish	<1 in 1,000,000	29%	1 in 1,400,000	
		Caucasian	1 in 410,000	83%	1 in 2,400,000	
		Latino	<1 in 1,000,000	74%	1 in 3,800,000	
		South Asian	<1 in 1,000,000	11%	1 in 1,100,000	
		Worldwide	1 in 770,000	74%	1 in 2,900,000	
Marfan Syndrome and Other *FBN1*-Related Disorders (AD) NM_000138.4	*FBN1*	Worldwide	1 in 5,000	85%	1 in 33,000	>95%
Medium Chain Acyl-CoA) Dehydrogenase Deficiency (AR) NM_000016.5	*ACADM*	African	1 in 120,000	69%	1 in 400,000	98%
		Ashkenazi Jewish	1 in 71,000	98%	1 in 3,600,000	
		East Asian	1 in 270,000	70%	1 in 930,000	
		Finnish	1 in 590,000	98%	1 in 29,600,000	
		Caucasian	1 in 13,000	94%	1 in 210,000	
		Latino	1 in 67,000	81%	1 in 350,000	
		South Asian	1 in 120,000	47%	1 in 230,000	
		Worldwide	1 in 31,000	86%	1 in 220,000	

Patient:		DOB:	8/21/2019	Lab #:	19040699PN

Metachromatic Leukodystrophy (AR)	*ARSA*	African	1 in 230,000	64%	1 in 640,000	98%
NM_000487.5		Ashkenazi Jewish	<1 in 1,000,000	67%	1 in 3,100,000	
		East Asian	1 in 530,000	74%	1 in 2,100,000	
		Finnish	1 in 270,000	94%	1 in 4,100,000	
		Caucasian	1 in 68,000	76%	1 in 280,000	
		Latino	<1 in 1,000,000	81%	1 in 5,300,000	
		South Asian	1 in 550,000	67%	1 in 1,700,000	
		Worldwide	1 in 130,000	74%	1 in 500,000	
		Sephardic Jewish - Yemenite	1 in 8,000	>90%	1 in 80,000	
		Navajo	1 in 2,500	>90%	1 in 25,000	
3-Methylcrotonyl-CoA Carboxylase Deficiency (*MCCC1*-Related) (AR)	*MCCC1*	African	1 in 280,000	26%	1 in 380,000	98%
NM_020166.4		East Asian	1 in 170,000	61%	1 in 430,000	
		Caucasian	1 in 500,000	81%	1 in 2,600,000	
		Latino	1 in 950,000	82%	1 in 5,300,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	1 in 710,000	69%	1 in 2,300,000	
3-Methylcrotonyl-CoA Carboxylase Deficiency (*MCCC2*-Related) (AR)	*MCCC2*	African	1 in 660,000	66%	1 in 1,900,000	98%
NM_022132.4		Ashkenazi Jewish	1 in 290,00	98%	1 in 14,300,000	
		East Asian	1 in 150,00	38%	1 in 240,000	
		Finnish	<1 in 1,000,000	63	1 in 2,700,000	
		Caucasian	1 in 170,000	%	1 in 530,000	
		Latino	in 63,000	95%	1 in 1,300,000	
		South Asian	1 i 0,000	48%	1 in 730,000	
		Worldwide	1 in 1 000	70%	1 in 600,000	
Methionine Adenosyltransferase I/III Deficiency (AR)	*MAT1A*	African	<1 in 1,000,	47%	1 in 1,900,000	98%
NM_000429.2		East Asian	1 in 1,000,000	29%	1 in 1,400,000	
		Caucasian	<1 in 1,000,000	23%	1 in 1,300,000	
		Latin	<1 in 1,000,000	63%	1 in 2,700,000	
		South As n	1 in 1,000,000	47%	1 in 1,900,000	
		Worldwide	<1 in 1,000,000	34%	1 in 1,500,000	
Methylmalonic Acidemia (*MMAA*-Related) (AR)	*MMAA*	East Asian	<1 in 1,000,000	98%	1 in 50,300,000	98%
NM_172250.2		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	91%	1 in 11,100,000	
		Latin	<1 in 1,000,000	75%	1 in 4,000,000	
		South Asian	<1 in 1,000,000	84%	1 in 6,100,000	
		Worldwide	<1 in 1,000,000	90%	1 in 9,500,000	
Methylmalonic Acidemia (*MMAB*-Related) (AR)	*MMAB*	African	<1 in 1,000,000	32%	1 in 1,500,000	98%
NM_052845.3		Finnish	<1 in 1,000,000	43%	1 in 1,800,000	
		Caucasian	<1 in 1,000,000	89%	1 in 9,100,000	
		Latino	<1 in 1,000,000	17%	1 in 1,200,000	
		South Asian	<1 in 1,000,000	24%	1 in 1,300,000	
		Worldwide	<1 in 1,000,000	59%	1 in 2,500,000	
Methylmalonic Acidemia (*MUT*-Related) (AR)	*MUT*	African	1 in 110,000	78%	1 in 510,000	98%
NM_000255.3		Ashkenazi Jewish	1 in 430,000	98%	1 in 21,700,000	
		East Asian	1 in 140,000	60%	1 in 360,000	
		Finnish	<1 in 1,000,000	74%	1 in 3,800,000	
		Caucasian	1 in 350,000	59%	1 in 860,000	
		Latino	1 in 150,000	91%	1 in 1,800,000	
		South Asian	1 in 280,000	62%	1 in 730,000	
		Worldwide	1 in 250,000	70%	1 in 840,000	
Methylmalonic Aciduria and, Homocystinuria Cobalamin C Type (AR)	*MMACHC*	African	1 in 310,000	89%	1 in 2,900,000	98%
NM_015506.2		Ashkenazi Jewish	1 in 160,000	98%	1 in 8,300,000	
		East Asian	1 in 140,000	75%	1 in 530,000	
		Caucasian	1 in 120,000	95%	1 in 2,400,000	
		Latino	1 in 42,000	98%	1 in 2,100,000	
		South Asian	1 in 210,000	76%	1 in 880,000	
		Worldwide	1 in 130,000	92%	1 in 1,700,000	

Patient:		DOB:	8/21/2019	Lab #:	19040699PN

Methylmalonic Aciduria and Homocystinuria, Cobalamin D Type (AR) NM_015702.2	*MMADHC*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	98%	1 in 50,300,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	98%	1 in 50,300,000	
Methylmalonic Aciduria and Homocystinuria, Cobalamin F Type (AR) NM_018368.3	*LMBRD1*	African	1 in 510,000	74%	1 in 1,900,000	90%
		East Asian	<1 in 1,000,000	90%	1 in 10,300,000	
		Finnish	<1 in 1,000,000	90%	1 in 10,300,000	
		Caucasian	1 in 680,000	88%	1 in 5,600,000	
		Latino	<1 in 1,000,000	90%	1 in 10,300,000	
		South Asian	<1 in 1,000,000	90%	1 in 10,300,000	
		Worldwide	<1 in 1,000,000	87%	1 in 7,800,000	
Methylmalonyl-CoA Epimerase Deficiency NM_032601.3	*MCEE*	Caucasian	<1 in 1,000,000	98%	1 in 50,300,000	98%
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	98%	1 in 50,300,000	
Mitochondrial Trifunctional Protein Deficiency (*HADHB*-Related) (AR) NM_000183.2	*HADHB*	African	<1 in 1,000,000	67%	1 in 3,000,000	90%
		East Asian	<1 in 1,000,000	37%	1 in 1,600,000	
		Finnish	<1 in 1,000,000	36%	1 in 1,600,000	
		Caucasian	<1 in 1,000,000	6[illegible]%	1 in 2,600,000	
		Latino	<1 in 1,000,000	[illegible]2%	1 in 5,600,000	
		South Asian	<1 in 1,000,000	75%	1 in 3,900,000	
		Worldwide	<1 in 1,000,000	62%	1 in 2,600,000	
Mucolipidosis II / IIIA (AR) NM_024312.4	*GNPTAB*	African	1 in 430,000	98%	1 in 21,600,000	98%
		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		East Asian	1 in 540,000	46%	1 in 1,000,000	
		Finnish	1 in 100,000	98%	1 in 5,100,000	
		Caucasian	1 in 200,000	80%	1 in 970,000	
		Latino	1 in 330,000	82%	1 in 1,800,000	
		South Asian	1 in 410,000	94%	1 in 6,800,000	
		Worldwide	1 in 24,000	83%	1 in 1,500,000	
Mucopolysaccharidosis Type I (AR) NM_000203.4	*IDUA*	African	1 in 560,000	82%	1 in 3,100,000	98%
		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		East Asian	1 in 220,000	40%	1 in 370,000	
		Finnish	1 in 140,000	98%	1 in 6,800,000	
		Caucasian	1 in 53,000	93%	1 in 780,000	
		Latino	1 in 690,000	84%	1 in 4,300,000	
		South Asian	1 in 52,000	95%	1 in 950,000	
		Worldwide	1 in 83,000	90%	1 in 810,000	
Mucopolysaccharidosis, Type II (XL) NM_000202.6 *Exception:* Exon 3	*IDS*	Worldwide	1 in 50,000	67%	1 in 150,000	90%
Mucopolysaccharidosis Type IVa (AR) NM_000512.4	*GALNS*	African	1 in 470,000	33%	1 in 700,000	92%
		Ashkenazi Jewish	<1 in 1,000,000	52%	1 in 2,100,000	
		East Asian	1 in 330,000	13%	1 in 370,000	
		Finnish	<1 in 1,000,000	74%	1 in 3,900,000	
		Caucasian	1 in 260,000	39%	1 in 430,000	
		Latino	1 in 440,000	54%	1 in 960,000	
		South Asian	1 in 440,000	14%	1 in 520,000	
		Worldwide	1 in 310,000	41%	1 in 520,000	
Mucopolysaccharidosis Type VI (AR) NM_000046.3	*ARSB*	African	<1 in 1,000,000	33%	1 in 1,500,000	98%
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	<1 in 1,000,000	73%	1 in 3,600,000	
		Caucasian	1 in 390,000	57%	1 in 910,000	
		Latino	<1 in 1,000,000	55%	1 in 2,200,000	
		South Asian	<1 in 1,000,000	72%	1 in 3,600,000	
		Worldwide	<1 in 1,000,000	54%	1 in 2,200,000	

Patient:		DOB:	8/21/2019	Lab #:	19040699PN

N-Acetylglutamate Synthase Deficiency (AR)	*NAGS*	African	<1 in 1,000,000	70%	1 in 3,400,000	98%
NM_153006.2		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	51%	1 in 2,100,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	37%	1 in 1,600,000	
		Worldwide	<1 in 1,000,000	70%	1 in 3,300,000	
Nephrogenic Diabetes Insipidus (*AVPR2*-Related) / Nephrogenic Syndrome of Inappropriate Antidiuresis (XL)	*AVPR2*	Worldwide	1 in 320,000	66%	1 in 940,000	96%
NM_000054.4						
Nephrogenic Diabetes Insipidus, Type II (AD/AR)	*AQP2*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		East Asian	<1 in 1,000,000	83%	1 in 6,000,000	
NM_000486.5		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	62%	1 in 2,700,000	
		Latino	1 in 840,000	93%	1 in 11,600,000	
		South Asian	<1 in 1,000,000	35%	1 in 1,500,000	
		Worldwide	<1 in 1,000,000	75%	1 in 4,000,000	
Neurodegeneration due to Cerebral Folate Transport Deficiency (AR)	*FOLR1*	African	<1 in 1,000,000	15%	1 in 1,200,000	72%
		Finnish	1 in 140,000	4[illegible]	1 in 280,000	
NM_016725.2		Caucasian	<1 in 1,000,000	[illegible]2%	1 in 2,600,000	
		South Asian	<1 in 1,000,000	41%	1 in 1,700,000	
		Worldwide	<1 in 1,000,000	54%	1 in 2,200,000	
Neuronal Ceroid-Lipofuscinosis (*CLN3*-Related) (AR)	*CLN3*	African	<1 in 1,000,000	59%	1 in 2,500,000	98%
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
NM_000089.2		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	1 in 240,000	95%	1 in 4,500,000	
		Latino	<1 in 1,000,000	51%	1 in 2,000,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	1 in 750,000	93%	1 in 10,300,000	
Neuronal Ceroid-Lipofuscinosis (*CLN5*-Related) (AR)	*CLN5*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
NM_006493.2		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	68%	1 in 3,100,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	46%	1 in 1,900,000	
		Worldwide	<1 in 1,000,000	80%	1 in 5,100,000	
Neuronal Ceroid-Lipofuscinosis (*CLN6*-Related) (AR)	*CLN6*	African	<1 in 1,000,000	63%	1 in 2,700,000	98%
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
NM_017882.2		Caucasian	<1 in 1,000,000	78%	1 in 4,600,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	89%	1 in 9,000,000	
		Worldwide	<1 in 1,000,000	85%	1 in 6,700,000	
Neuronal Ceroid-Lipofuscinosis (*CLN8*-Related) (AR)	*CLN8*	African	<1 in 1,000,000	31%	1 in 1,500,000	98%
		East Asian	<1 in 1,000,000	25%	1 in 1,300,000	
NM_018941.3		Finnish	1 in 630,000	85%	1 in 4,100,000	
		Caucasian	<1 in 1,000,000	33%	1 in 1,500,000	
		Latino	<1 in 1,000,000	44%	1 in 1,800,000	
		South Asian	<1 in 1,000,000	55%	1 in 2,200,000	
		Worldwide	<1 in 1,000,000	52%	1 in 2,100,000	
Neuronal Ceroid-Lipofuscinosis (*MFSD8*-Related) (AR)	*MFSD8*	African	<1 in 1,000,000	66%	1 in 3,000,000	98%
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
NM_152778.2		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	83%	1 in 5,900,000	
		Latino	<1 in 1,000,000	70%	1 in 3,400,000	
		South Asian	1 in 920,000	86%	1 in 6,700,000	
		Worldwide	<1 in 1,000,000	86%	1 in 7,100,000	

Patient:		DOB:	8/21/2019	Lab #:	19040699PN

Neuronal Ceroid-Lipofuscinosis (*PPT1*-Related) (AR) NM_000310.3	*PPT1*	African	<1 in 1,000,000	83%	1 in 5,700,000	98%
		East Asian	<1 in 1,000,000	31%	1 in 1,500,000	
		Finnish	1 in 22,000	98%	1 in 1,100,000	
		Caucasian	1 in 290,000	93%	1 in 4,100,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	90%	1 in 10,000,000	
		Worldwide	1 in 320,000	93%	1 in 4,600,000	
Neuronal Ceroid-Lipofuscinosis (*TPP1*-Related) (AR) NM_000391.3	*TPP1*	African	<1 in 1,000,000	36%	1 in 1,600,000	98%
		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		East Asian	<1 in 1,000,000	26%	1 in 1,400,000	
		Finnish	1 in 500,000	98%	1 in 25,200,000	
		Caucasian	1 in 280,000	92%	1 in 3,400,000	
		Latino	<1 in 1,000,000	79%	1 in 4,800,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	1 in 570,000	87%	1 in 4,500,000	
Niemann-Pick Disease (*SMPD1*-Related) (AR) NM_000543.4	*SMPD1*	African	1 in 57,000	80%	1 in 290,000	98%
		Ashkenazi Jewish	1 in 39,000	98%	1 in 1,900,000	
		East Asian	1 in 26,00	88%	1 in 220,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	1 in 490,000	%	1 in 1,400,000	
		Latino	n 1,000,000	76%	1 in 4,200,000	
		South Asian	1 30,000	57%	1 in 1,000,000	
		Worldwide	1 in 2 000	77%	1 in 990,000	
Omenn Syndrome (*RAG2*-Related) (AR) NM_000536.2	*RAG2*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		Ashkenazi Jewi	1 in 1,000,000	98%	1 in 50,300,000	
		Finnis	<1 in 1,000,000	98%	1 in 50,300,000	
		Cauca	<1 in 1,000,000	73%	1 in 3,700,000	
		South As	<1 in 1,000,000	6%	1 in 1,100,000	
		Worldwid	<1 in 1,000,000	66%	1 in 2,900,000	
Omenn Syndrome / Severe Combined Immunodeficiency, Athabaskan-Type (AR) NM_001033855.1	*DCLRE1C*	rican	<1 in 1,000,000	88%	1 in 8,400,000	96%
		st A	<1 in 1,000,000	96%	1 in 25,300,000	
		nish	<1 in 1,000,000	58%	1 in 2,400,000	
		Cauca n	<1 in 1,000,000	70%	1 in 3,300,000	
		tino	<1 in 1,000,000	75%	1 in 4,100,000	
		South Asian	<1 in 1,000,000	48%	1 in 1,900,000	
		Worldwide	<1 in 1,000,000	76%	1 in 4,200,000	
		Navajo and Apache Native American	1 in 9,000	>90%	1 in 90,000	
Omenn Syndrome and other *RAG1*-Related Disorders (AR) NM_000448.2	*RAG1*	African	1 in 610,000	46%	1 in 1,100,000	94%
		Ashkenazi Jewish	<1 in 1,000,000	23%	1 in 1,300,000	
		East Asian	1 in 62,000	10%	1 in 69,000	
		Finnish	<1 in 1,000,000	50%	1 in 2,000,000	
		Caucasian	1 in 420,000	38%	1 in 680,000	
		Latino	1 in 850,000	57%	1 in 2,000,000	
		South Asian	<1 in 1,000,000	33%	1 in 1,500,000	
		Worldwide	1 in 450,000	35%	1 in 690,000	
Ornithine Aminotransferase Deficiency (AR) NM_000274.3	*OAT*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	1 in 76,000	98%	1 in 3,800,000	
		Caucasian	<1 in 1,000,000	78%	1 in 4,600,000	
		Latino	<1 in 1,000,000	47%	1 in 1,900,000	
		South Asian	<1 in 1,000,000	57%	1 in 2,300,000	
		Worldwide	<1 in 1,000,000	83%	1 in 6,000,000	
		Sephardic Jewish - Iraqi and Syrian	1 in 125,000	>90%	1 in 1,300,000	
Ornithine Transcarbamylase Deficiency (XL) NM_000531.5	*OTC*	Worldwide	1 in 60,000	71%	1 in 210,000	99%

Patient:		DOB:	8/21/2019	Lab #:	19040699PN

Osteopetrosis 1 (AR) NM_006019.2	*TCIRG1*	African	1 in 700,000	76%	1 in 2,900,000	96%
		Ashkenazi Jewish	1 in 970,000	78%	1 in 4,400,000	
		East Asian	1 in 420,000	89%	1 in 3,800,000	
		Finnish	<1 in 1,000,000	96%	1 in 25,300,000	
		Caucasian	1 in 640,000	84%	1 in 3,900,000	
		Latino	1 in 690,000	96%	1 in 17,300,000	
		South Asian	<1 in 1,000,000	78%	1 in 4,600,000	
		Worldwide	1 in 640,000	87%	1 in 4,800,000	
		Costa Rican	1 in 30,000	>90%	1 in 300,000	
		Chuvashiyan	1 in 14,000	>90%	1 in 140,000	
Permanent Neonatal Diabetes Mellitus (*INS*-Related) (AD) NM_000207.2	*INS*	Worldwide	1 in 220,000	70%	1 in 730,000	97%
Phenylalanine Hydroxylase Deficiency (AR) NM_000277.1	*PAH*	African	1 in 82,000	74%	1 in 320,000	98%
		Ashkenazi Jewish	1 in 1,100	97%	1 in 41,000	
		East Asian	1 in 18,000	29%	1 in 26,000	
		Finnish	1 in 99,000	57%	1 in 230,000	
		Caucasian	1 in 5,60[illegible]	79%	1 in 27,000	
		Latino	1 in 20,000	76%	1 in 84,000	
		South Asian	1 in 59,000	66[illegible]	1 in 170,000	
		Worldwide	1 in 10,000	[illegible]%	1 in 43,000	
		Turkish	[illegible] in 4,000	40%	1 in 6,700	
		Irish	1 [illegible],000	83%	1 in 29,400	
		Sicilian	1 in [illegible]00	23%	1 in 3,900	
		Sephardic Jewish - Iranian, [illegible]ukharian, Kavkazi, Tunisian and [illegible]occan	1 in 1,30[illegible]	77%	1 in 5,600	
3-Phosphoglycerate Dehydrogenase Deficiency (AR) NM_006623.3	*PHGDH*	Afric[illegible]	<1 in 1,000,000	41%	1 in 1,700,000	98%
		Ashkenazi [illegible]h	1 in 360,000	98%	1 in 17,900,000	
		East Asi[illegible]	[illegible] in 1,000,000	98%	1 in 50,300,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Ca[illegible]sian	<1 in 1,000,000	98%	1 in 50,300,000	
		[illegible]atino	<1 in 1,000,000	47%	1 in 1,900,000	
		Sou[illegible] Asian	<1 in 1,000,000	60%	1 in 2,500,000	
		World[illegible]e	<1 in 1,000,000	89%	1 in 8,900,000	
Primary Carnitine Deficiency (AR) NM_003060.2	*SLC22A5*	[illegible]an	1 in 38,000	89%	1 in 340,000	96%
		Ashkenazi Jewish	<1 in 1,000,000	96%	1 in 25,300,000	
		East Asian	1 in 19,000	79%	1 in 88,000	
		Finnish	<1 in 1,000,000	65%	1 in 2,900,000	
		Caucasian	1 in 250,000	69%	1 in 800,000	
		Latino	1 in 290,000	74%	1 in 1,100,000	
		South Asian	1 in 11,000	92%	1 in 140,000	
		Worldwide	1 in 83,000	82%	1 in 460,000	
		Faroese	1 in 1,600	>90%	1 in 16,000	
Primary Hyperoxaluria, Type 1 (AR) NM_000030.2	*AGXT*	African	1 in 430,000	78%	1 in 1,900,000	98%
		Ashkenazi Jewish	<1 in 1,000,000	56%	1 in 2,300,000	
		East Asian	1 in 72,000	89%	1 in 660,000	
		Finnish	<1 in 1,000,000	66%	1 in 3,000,000	
		Caucasian	1 in 180,000	80%	1 in 860,000	
		Latino	1 in 690,000	73%	1 in 2,500,000	
		South Asian	1 in 370,000	70%	1 in 1,200,000	
		Worldwide	1 in 260,000	79%	1 in 1,200,000	
Primary Hyperoxaluria, Type 2 (AR) NM_012203.1	*GRHPR*	African	<1 in 1,000,000	43%	1 in 1,700,000	98%
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	1 in 750,000	92%	1 in 9,400,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	1 in 430,000	94%	1 in 7,000,000	
		Worldwide	1 in 960,000	86%	1 in 6,600,000	

Patient:		DOB:	8/21/2019	Lab #:	19040699PN

Primary Hyperoxaluria, Type 3 (AR)	*HOGA1*	African	1 in 640,000	92%	1 in 7,600,000	98%
NM_138413.3		Ashkenazi Jewish	1 in 5,400	98%	1 in 270,000	
		East Asian	1 in 59,000	98%	1 in 3,000,000	
		Finnish	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	1 in 110,000	87%	1 in 860,000	
		Latino	1 in 350,000	88%	1 in 2,900,000	
		South Asian	<1 in 1,000,000	80%	1 in 5,100,000	
		Worldwide	1 in 140,000	90%	1 in 1,400,000	
Propionic Acidemia (*PCCA*-Related) (AR)	*PCCA*	African	1 in 620,000	51%	1 in 1,300,000	86%
NM_000282.3		Ashkenazi Jewish	<1 in 1,000,000	68%	1 in 3,200,000	
		East Asian	1 in 700,000	70%	1 in 2,400,000	
		Finnish	<1 in 1,000,000	86%	1 in 7,400,000	
		Caucasian	<1 in 1,000,000	57%	1 in 2,300,000	
		Latino	1 in 740,000	35%	1 in 1,100,000	
		South Asian	<1 in 1,000,000	60%	1 in 2,500,000	
		Worldwide	1 in 970,000	50%	1 in 1,900,000	
Propionic Acidemia (*PCCB*-Related) (AR)	*PCCB*	African	1 in 270,000	92%	1 in 3,100,000	98%
NM_000532.4		East Asian	1 in 150,00	63%	1 in 400,000	
		Finnish	<1 in 1,000,0	80%	1 in 5,000,000	
		Caucasian	<1 in 1,000,000	9	1 in 9,900,000	
		Latino	<1 in 1,000,000	%	1 in 2,600,000	
		South Asian	<1 in 1,000,000	59%	1 in 2,400,000	
		Worldwide	<1 in ,000,000	80%	1 in 4,900,000	
Pyridoxamine 5'-Phosphate Oxidase Deficiency (AR)	*PNPO*	African	in 1, 000	92%	1 in 12,800,000	92%
		East Asian	<1 in 1,000,	72%	1 in 3,600,000	
NM_018129.3		Finnish	1 in 1,000,000	71%	1 in 3,500,000	
		Cauca n	<1 in 1,000,000	78%	1 in 4,500,000	
		Latin	<1 in 1,000,000	92%	1 in 12,800,000	
		South As n	1 in 1,000,000	59%	1 in 2,400,000	
		Worldwide	<1 in 1,000,000	78%	1 in 4,500,000	
Pyridoxine-Dependent Epilepsy (AR)	*ALDH7A1*	an	1 in 200,000	55%	1 in 440,000	92%
NM_001182.4		t Asi	1 in 290,000	56%	1 in 650,000	
		ish	<1 in 1,000,000	21%	1 in 1,300,000	
		Cauca n	1 in 380,000	53%	1 in 800,000	
		ino	1 in 780,000	74%	1 in 3,000,000	
		South Asian	1 in 870,000	41%	1 in 1,500,000	
		Worldwide	1 in 430,000	52%	1 in 900,000	
6-Pyruvoyl-Tetrahydropterin Synthase Deficiency (AR)	*PTS*	African	<1 in 1,000,000	98%	1 in 50,300,000	98%
		Ashkenazi Jewish	<1 in 1,000,000	98%	1 in 50,300,000	
NM_000317.2		East Asian	1 in 97,000	89%	1 in 910,000	
		Finnish	1 in 530,000	81%	1 in 2,700,000	
		Caucasian	1 in 920,000	55%	1 in 2,000,000	
		Latino	<1 in 1,000,000	65%	1 in 2,800,000	
		South Asian	1 in 470,000	70%	1 in 1,600,000	
		Worldwide	1 in 630,000	66%	1 in 1,800,000	
Retinoblastoma (AD)	*RB1*	Worldwide	1 in 20,000	80%	1 in 100,000	96%
NM_000321.2						
Segawa Syndrome (AR)	*TH*	African	<1 in 1,000,000	45%	1 in 1,800,000	98%
NM_000360.3		East Asian	1 in 370,000	80%	1 in 1,900,000	
		Caucasian	<1 in 1,000,000	74%	1 in 3,800,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	76%	1 in 4,200,000	
Sepiapterin Reductase Deficiency (AR)	*SPR*	Finnish	<1 in 1,000,000	92%	1 in 12,800,000	92%
NM_003124.4		Caucasian	<1 in 1,000,000	92%	1 in 12,800,000	
		Latino	<1 in 1,000,000	92%	1 in 12,800,000	
		South Asian	<1 in 1,000,000	92%	1 in 12,800,000	
		Worldwide	<1 in 1,000,000	92%	1 in 12,800,000	

Patient:		DOB:	8/21/2019	Lab #:	19040699PN

Severe Combined Immunodeficiency (*IL7R*-Related) (AR) NM_002185.3	*IL7R*	African	<1 in 1,000,000	88%	1 in 8,600,000	88%
		East Asian	<1 in 1,000,000	88%	1 in 8,600,000	
		Caucasian	<1 in 1,000,000	88%	1 in 8,600,000	
		Latino	<1 in 1,000,000	88%	1 in 8,600,000	
		South Asian	<1 in 1,000,000	88%	1 in 8,600,000	
		Worldwide	<1 in 1,000,000	88%	1 in 8,600,000	
Severe Combined Immunodeficiency (*JAK3*-Related) (AR) NM_000215.3	*JAK3*	African	<1 in 1,000,000	96%	1 in 25,300,000	96%
		Caucasian	<1 in 1,000,000	21%	1 in 1,300,000	
		Latino	<1 in 1,000,000	26%	1 in 1,300,000	
		South Asian	<1 in 1,000,000	54%	1 in 2,200,000	
		Worldwide	<1 in 1,000,000	52%	1 in 2,100,000	
Severe Combined Immunodeficiency (*PTPRC*-Related) (AR)	*PTPRC*	East Asian	1 in 860,000	64%	1 in 2,400,000	64%
		Caucasian	<1 in 1,000,000	64%	1 in 2,800,000	
		Latino	<1 in 1,000,000	64%	1 in 2,800,000	
		Worldwide	<1 in 1,000,000	64%	1 in 2,800,000	
Severe Neonatal Hyperparathyroidism / Autosomal Dominant Hypocalcemia) (AD/AR) NM_000388.3	*CASR*	African	<1 in 1,000,000	33%	1 in 1,500,000	98%
		East Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Caucasian	<1 in 1,000,000	13%	1 in 1,200,000	
		South Asian	<1 in 1,000,000	55%	1 in 2,200,000	
		Worldwide	**<1 in 1,000,000**	**26%**	**1 in 1,400,000**	
Spherocytosis, Type 1 (AD) NM_000037.3	*ANK1*	Worldwide	1 in 2,000	2%	1 in 11,000	99%
Spherocytosis, Type 5 (AR) NM_000119.2	*EPB42*	African	1 in 20,000	60%	1 in 2,000,000	86%
		Ashkenazi Jewish	1 in 30,000	86%	1 in 220,000	
		East Asian	1 in 690,000	45%	1 in 1,300,000	
		Finnish	1 in 690,000	86%	1 in 5,100,000	
		Caucasian	<1 in 1,000,000	57%	1 in 2,300,000	
		Latino	<1 in 1,000,000	37%	1 in 1,600,000	
		South Asian	<1 in 1,000,000	86%	1 in 7,400,000	
		Worldwide	1 in 680,000	71%	1 in 2,300,000	
Spinal Muscular Atrophy (AR) NM_000344.3	*SMN1*	African	1 in 17,000	>95%	1 in 440,000	>95%
		Ashkenazi Jewish	1 in 7,000	>95%	1 in 140,000	
		Asian	1 in 11,000	>95%	1 in 220,000	
		Caucasian	1 in 5,000	>95%	1 in 100,000	
		Latino	1 in 55,000	>95%	1 in 1,100,000	
		Worldwide	1 in 10,000	>95%	1 in 200,000	
Tay-Sachs Disease (AR) NM_000520.4	*HEXA*	African	1 in 190,000	98%	1 in 9,300,000	98%
		Ashkenazi Jewish	1 in 3,600	98%	1 in 180,000	
		East Asian	1 in 180,000	98%	1 in 8,900,000	
		Finnish	1 in 640,000	98%	1 in 32,000,000	
		Caucasian	1 in 33,000	95%	1 in 640,000	
		Latino	1 in 240,000	79%	1 in 1,100,000	
		South Asian	1 in 690,000	48%	1 in 1,300,000	
		Worldwide	1 in 59,000	93%	1 in 800,000	
Thyroid Dyshormonogenesis 1 (AR) NM_000453.2	*SLC5A5*	African	<1 in 1,000,000	88%	1 in 8,600,000	88%
		Ashkenazi Jewish	1 in 800,000	88%	1 in 6,900,000	
		East Asian	<1 in 1,000,000	57%	1 in 2,300,000	
		Caucasian	<1 in 1,000,000	88%	1 in 8,600,000	
		Latino	1 in 270,000	88%	1 in 2,300,000	
		South Asian	<1 in 1,000,000	88%	1 in 8,600,000	
		Worldwide	<1 in 1,000,000	83%	1 in 6,000,000	
Thyroid Dyshormonogenesis 2A (AR) NM_000547.5	*TPO*	African	1 in 160,000	25%	1 in 210,000	96%
		Ashkenazi Jewish	1 in 160,000	30%	1 in 220,000	
		East Asian	1 in 13,000	71%	1 in 43,000	
		Finnish	1 in 160,000	43%	1 in 290,000	
		Caucasian	1 in 140,000	63%	1 in 380,000	
		Latino	1 in 150,000	57%	1 in 350,000	
		South Asian	<1 in 1,000,000	18%	1 in 1,200,000	
		Worldwide	1 in 120,000	54%	1 in 250,000	

Thyroid Dyshormonogenesis 3 (AR)	*TG*	African	1 in 65,000	75%	1 in 260,000	96%
NM_003235.4		Ashkenazi Jewish	1 in 81,000	91%	1 in 870,000	
		East Asian	1 in 17,000	24%	1 in 22,000	
		Finnish	1 in 220,000	53%	1 in 470,000	
		Caucasian	1 in 120,000	64%	1 in 330,000	
		Latino	1 in 91,000	91%	1 in 1,000,000	
		South Asian	<1 in 1,000,000	82%	1 in 5,500,000	
		Worldwide	1 in 89,000	59%	1 in 220,000	
Thyroid Dyshormonogenesis 4 (AR)	*IYD*	African	1 in 910,000	83%	1 in 5,400,000	98%
NM_203395.2		Caucasian	<1 in 1,000,000	44%	1 in 1,800,000	
		Latino	<1 in 1,000,000	33%	1 in 1,500,000	
		South Asian	<1 in 1,000,000	18%	1 in 1,200,000	
		Worldwide	<1 in 1,000,000	57%	1 in 2,300,000	
Thyroid Dyshormonogenesis 5 (AR)	*DUOXA2*	African	1 in 810,000	88%	1 in 6,900,000	88%
NM_207581.3		East Asian	1 in 33,000	87%	1 in 240,000	
		Caucasian	<1 in 1,000,000	88%	1 in 8,600,000	
		Latino	<1 in 1,000,000	88%	1 in 8,600,000	
		South Asian	<1 in 1,000,000	88%	1 in 8,600,000	
		Worldwide	<1 in 1,000,000	87%	1 in 8,000,000	
Thyroid Dyshormonogenesis 6 (AR)	*DUOX2*	African	1 in 31,000	67%	1 in 95,000	98%
NM_207581.3		Ashkenazi Jewish	<1 in 1,000,000	2%	1 in 2,600,000	
		East Asian	1 in 500	45%	1 in 900	
		Finnish	1 in 6,000	87%	1 in 46,000	
		Caucasian	1 in 1,000	48%	1 in 28,000	
		Latino	1 in 42,000	54%	1 in 91,000	
		South Asian	1 in 1,000,000	18%	1 in 1,200,000	
		Worldwide	1 in 9,900	53%	1 in 21,000	
Exception: Exons 6 and 7						
Tyrosinemia, Type I (AR)	*FAH*	African	1 in 510,000	83%	1 in 2,900,000	98%
NM_000137.2		Ashkenazi Jewish	1 in 71,000	98%	1 in 3,600,000	
		East Asian	<1 in 1,000,000	15%	1 in 1,200,000	
		Finnish	1 in 420,000	98%	1 in 20,900,000	
		Caucasian	1 in 270,000	74%	1 in 1,000,000	
		Latino	<1 in 1,000,000	98%	1 in 50,300,000	
		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	1 in 410,000	79%	1 in 1,900,000	
		French Canadian - Saguenay	1 in 2,500	>90%	1 in 25,000	
		Lac-St. Jean French Canadian - Other	1 in 17,000	>90%	1 in 170,000	
Tyrosinemia, Type II (AR)	*TAT*	African	<1 in 1,000,000	88%	1 in 8,600,000	88%
NM_000353.2		East Asian	<1 in 1,000,000	10%	1 in 1,100,000	
		Caucasian	<1 in 1,000,000	22%	1 in 1,300,000	
		South Asian	<1 in 1,000,000	53%	1 in 2,100,000	
		Worldwide	<1 in 1,000,000	41%	1 in 1,700,000	
Tyrosinemia, Type III (AR)	*HPD*	Caucasian	<1 in 1,000,000	98%	1 in 50,300,000	98%
NM_002150.2		South Asian	<1 in 1,000,000	98%	1 in 50,300,000	
		Worldwide	<1 in 1,000,000	68%	1 in 3,200,000	
Very Long Chain Acyl-CoA Dehydrogenase Deficiency (AR)	*ACADVL*	African	1 in 85,000	57%	1 in 200,000	96%
NM_000018.3		Ashkenazi Jewish	<1 in 1,000,000	53%	1 in 2,100,000	
		East Asian	1 in 160,000	22%	1 in 210,000	
		Finnish	1 in 340,000	87%	1 in 2,700,000	
		Caucasian	1 in 48,000	78%	1 in 220,000	
		Latino	1 in 290,000	45%	1 in 520,000	
		South Asian	1 in 550,000	52%	1 in 1,100,000	
		Worldwide	1 in 97,000	69%	1 in 310,000	
Wilms Tumor, Type 1 and Other *WT1*-Related Disorders (AD)	*WT1*	Worldwide	1 in 200,000	79%	1 in 950,000	99%
NM_024426.4						

Patient:		DOB:	8/21/2019	Lab #:	19040699PN

Wolman Disease / Cholesteryl Ester Storage Disease (AR) NM_000235.3	*LIPA*	African	<1 in 1,000,000	71%	1 in 3,400,000	96%
		Ashkenazi Jewish	<1 in 1,000,000	96%	1 in 25,300,000	
		East Asian	<1 in 1,000,000	96%	1 in 25,300,000	
		Finnish	<1 in 1,000,000	60%	1 in 2,500,000	
		Caucasian	1 in 220,000	86%	1 in 1,600,000	
		Latino	1 in 430,000	72%	1 in 1,500,000	
		South Asian	<1 in 1,000,000	96%	1 in 25,300,000	
		Worldwide	1 in 430,000	84%	1 in 2,700,000	
		Sephardic Jewish - Iranian	1 in 2,700	>90%	1 in 27,000	
X-Linked Severe Combined Immunodeficiency (XL) NM_000206.2	*IL2RG*	Worldwide	1 in 50,000	90%	1 in 500,000	99%

AD: Autosomal Dominant
AR: Autosomal Recessive
XL: X-Linked