Alabama Fertility

Records Release

ARIEL JENNIS~Hey Ariel! I hope this finds you well! Please complete and return to me so I can release records to you. You can email back to mdurham@alabamafertility.com or fax 256-533-5263. Once received, I'll send your records to you! Let me know if you have any questions. Talk to you soon! Mary D.

Please send my health information to:

To/From: Alabama Fertility Specialists

3490 Independence Drive
Birmingham, Alabama 35209
Telephone (256) 533-1010
Office Fax (256) 533-5263

To/From: Ian Kramer
iankramer@ymail.com

Fax:

Items needed: All records.

Purpose of release: Records

I understand that:

1. This authorization is voluntary. I may refuse to sign this authorization and my treatment and/or payment obligations will not be affected.
2. This authorization will remain in effect for one (1) year or until I revoke it in writing, which I may do at any time.
3. My records may contain hepatitis and HIV screening results that may be included in the record release.
4. The sender of this health information cannot guarantee that the recipient of the information will not re-disclose this information.
5. I have the right to receive a copy of this authorization form after I sign it.

Patient Name: Kramer (Last) Ian (First) A (MI)

Social Security #: 148.94.0723 Date of Birth: 02/19/1993

Telephone #: (862) 266-6360

Patient Signature: [signature] Date: 11/01/24

Alabama Fertility

, Alabama Fertility
3490 Independence Drive
Homewood, AL 35209

KRAMER, IAN
DOB: , 02/19/93
ACCOUNT #: CL-257AJ-1777666

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Infectious disease

Anti-HCV - Partner	11/15/23	Negative:0.068 (IAN, KRAMER)
HBs Ag - Partner	11/15/23	Negative:0.366 (IAN, KRAMER)
HIV1-2 Ab/Ag - Partner	11/15/23	Negative (IAN, KRAMER)
Syphilis - Partner	11/15/23	Negative:0.129 (IAN, KRAMER)
GC/CT- urine - Partner	11/15/23	Negative (IAN, KRAMER)
Anti-HCV - Partner	11/29/22	Negative:0.050 (IAN, KRAMER)
HBs Ag - Partner	11/29/22	Negative:0.391 (IAN, KRAMER)
Syphilis - Partner	11/29/22	Negative:0.109 (IAN, KRAMER)
HIV1-2 Ab/Ag - Partner	11/29/22	Negative (IAN, KRAMER)
GC/CT- urine - Partner	11/29/22	Negative (IAN, KRAMER)

Other

Expanded Carrier Screen (283) (S4+279 genes) - Partner •	11/29/22	Carrier CPT, otherwise negative (IAN, KRAMER)

Patient Information
Name: Ian Kramer
Date of Birth: 02/19/1993
Sema4 ID: 22241019
Client ID: 400534
Indication: Carrier Screening

Specimen Information
Specimen Type: Blood
Date Collected: 11/29/2022
Date Received: 11/30/2022
Final Report: 12/20/2022

Referring Provider
Merry Lynn Mann, M.D.
Alabama Fertility
3490 Independence Drive
Birmingham, AL. 35209
Fax: 205-874-7021

Expanded Carrier Screen (283 genes)

with Personalized Residual Risk

SUMMARY OF RESULTS AND RECOMMENDATIONS

⊕ Positive	⊖ Negative
Carrier of Carnitine Palmitoyltransferase II Deficiency (AR) Associated gene(s): *CPT2* Variant(s) Detected: c.1798G>C, p.G600R, Likely Pathogenic, Hétérozygous (one copy)	**Negative for all other genes tested** To view a full list of genes and diseases tested please see Table 1 in this report

AR=Autosomal recessive; XL=X-linked

Recommendations

- Testing the partner for the above positive disorder(s) and genetic counseling are recommended.
- Please note that for female carriers of X-linked diseases, follow-up testing of a male partner is not indicated.
- CGG repeat analysis of *FMR1* for fragile X syndrome is not performed on males as repeat expansion of premutation alleles is not expected in the male germline.
- Individuals of Asian, African, Hispanic and Mediterranean ancestry should also be screened for hemoglobinopathies by CBC and hemoglobin electrophoresis.
- Consideration of residual risk by ethnicity after a negative carrier screen is recommended for the other diseases on the panel, especially in the case of a positive family history for a specific disorder. Please note that residual risks for X-linked diseases (including full repeat expansions for Fragile X syndrome) may not be accurate for males and the actual residual risk is likely to be lower.
- As genetic technologies may improve and variant classifications may change over time, it is recommended to obtain a new carrier screening test or reanalysis when a new pregnancy is being considered.

Interpretation of positive results

Carnitine Palmitoyltransferase II Deficiency (AR)

Results and Interpretation

A heterozygous (one copy) likely pathogenic missense variant, c.1798G>C, p.G600R, was detected in the *CPT2* gene (NM_000098.2). When this variant is present in trans with a pathogenic variant, it is considered to be causative for carnitine palmitoyltransferase II deficiency. Therefore, this individual is expected to be at least a carrier for carnitine palmitoyltransferase II deficiency. Heterozygous carriers are not expected to exhibit symptoms of this disease.

What is Carnitine Palmitoyltransferase II Deficiency?

Carnitine palmitoyltransferase II deficiency is an autosomal recessive disorder caused by pathogenic variants in the gene *CPT2*. While it is diagnosed in individuals worldwide, it has a higher prevalence among individuals of Ashkenazi Jewish descent. There are three forms of carnitine palmitoyltransferase II deficiency: (a) the lethal neonatal form, (b) the severe infantile hepatocardiomuscular form, and (c) the

myopathic form. Both the lethal neonatal form and severe infantile hepatocardiomuscular form are severe multisystemic diseases. Symptoms include liver failure with hypoketotic hypoglycemia, cardiomyopathy, cardiac arrhythmias, seizures, and early death. These symptoms are present shortly after birth or within the first year of life. The myopathic form presents between the first to sixth decade of life and includes symptoms of muscle pain and weakness during periods of prolonged exercise, cold exposure, or stress. Specific variants have been associated with the different forms of the disease, and therefore it may be possible to predict the phenotype in some patients.

Test description

This patient was tested for a panel of diseases using a combination of sequencing, targeted genotyping and copy number analysis. Please note that negative results reduce but do not eliminate the possibility that this individual is a carrier for one or more of the disorders tested. Please see Table 1 for a list of genes and diseases tested with the patient's personalized residual risk. If personalized residual risk is not provided, please see the complete residual risk table at **go.sema4.com/residualrisk**. Only variants determined to be pathogenic or likely pathogenic are reported in this carrier screening test.

Preti Jain

Preti Jain, Ph.D., FACMG, DABMGG, Director - Molecular Genetics

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Carrier screening report
Ian Kramer
Date of Birth: 02/19/1993
Sema4 ID: 22241019

Genes and diseases tested

The personalized residual risks listed below are specific to this individual. The complete residual risk table is available at **go.sema4.com/residualrisk**

Table 1: List of genes and diseases tested with detailed results

Disease	Gene	Inheritance Pattern	Status	Detailed Summary
Positive				
Carnitine Palmitoyltransferase II Deficiency	*CPT2*	AR	Carrier	c.1798G>C, p.G600R, Likely Pathogenic. Heterozygous (one copy)
Negative				
3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency	*HSD3B2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,300
3-Methylcrotonyl-CoA Carboxylase Deficiency (*MCCC1*-Related)	*MCCC1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,500
3-Methylcrotonyl-CoA Carboxylase Deficiency (*MCCC2*-Related)	*MCCC2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,200
3-Methylglutaconic Aciduria, Type III	*OPA3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 29,000
3-Phosphoglycerate Dehydrogenase Deficiency	*PHGDH*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 30,000
6-Pyruvoyl-Tetrahydropterin Synthase Deficiency	*PTS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,800
***WNT10A*-Related Ectodermal Dysplasia**	*WNT10A*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,900
Abetalipoproteinemia	*MTTP*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,200
Achromatopsia (*CNGB3*-related)	*CNGB3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 8,600
Acrodermatitis Enteropathica	*SLC39A4*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 12,000
Acute Infantile Liver Failure	*TRMU*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 9,400
Acyl-CoA Oxidase I Deficiency	*ACOX1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 39,000
Adenosine Deaminase Deficiency	*ADA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,300
Adrenoleukodystrophy, X-Linked	*ABCD1*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 19,000
Aicardi-Goutieres Syndrome (*SAMHD1*-Related)	*SAMHD1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 10,000
Alpha-Mannosidosis	*MAN2B1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,200
Alpha-Thalassemia	*HBA1/HBA2*	AR	Reduced Risk	*HBA1* Copy Number: 2 *HBA2* Copy Number: 2 No pathogenic copy number variants detected *HBA1/HBA2* Sequencing: Negative **Personalized Residual Risk:** 1 in 490
Alpha-Thalassemia Intellectual Disability Syndrome	*ATRX*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 48,000
Alport Syndrome (*COL4A3*-Related)	*COL4A3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,800
Alport Syndrome (*COL4A4*-Related)	*COL4A4*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,800
Alport Syndrome (*COL4A5*-Related)	*COL4A5*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 150,000
Alstrom Syndrome	*ALMS1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,800
Andermann Syndrome	*SLC12A6*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 151,000
Argininosuccinic Aciduria	*ASL*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,200
Aromatase Deficiency	*CYP19A1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,400
Arthrogryposis, Intellectual Disability, and Seizures	*SLC35A3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 37,000
Asparagine Synthetase Deficiency	*ASNS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 21,000
Aspartylglycosaminuria	*AGA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 13,000
Ataxia With Isolated Vitamin E Deficiency	*TTPA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 61,000
Ataxia-Telangiectasia	*ATM*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,300

Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	*SACS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,600
Bardet-Biedl Syndrome (*BBS10*-Related)	*BBS10*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,700
Bardet-Biedl Syndrome (*BBS12*-Related)	*BBS12*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 9,900
Bardet-Biedl Syndrome (*BBS1*-Related)	*BBS1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,400
Bardet-Biedl Syndrome (*BBS2*-Related)	*BBS2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,200
Bare Lymphocyte Syndrome, Type II	*CIITA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 35,000
Bartter Syndrome, Type 4A	*BSND*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 91,000
Bernard-Soulier Syndrome, Type A1	*GP1BA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 42,000
Bernard-Soulier Syndrome, Type C	*GP9*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,100
Beta-Globin-Related Hemoglobinopathies	*HBB*	AR	Reduced Risk	**Personalized Residual Risk (Beta-Globin-Related Hemoglobinopathies):** 1 in 2,000 **Personalized Residual Risk (Beta-Globin-Related Hemoglobinopathies: HbS Variant):** 1 in 11,000 **Personalized Residual Risk (Beta-Globin-Related Hemoglobinopathies: HbC Variant):** 1 in 42,000
Beta-Ketothiolase Deficiency	*ACAT1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,400
Bilateral Frontoparietal Polymicrogyria	*GPR56*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 62,000
Biotinidase Deficiency	*BTD*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 500
Bloom Syndrome	*BLM*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 7,400
Canavan Disease	*ASPA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,400
Carbamoylphosphate Synthetase I Deficiency	*CPS1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,100
Carnitine Palmitoyltransferase IA Deficiency	*CPT1A*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 24,000
Carpenter Syndrome	*RAB23*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 21,000
Cartilage-Hair Hypoplasia	*RMRP*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 960
Cerebral Creatine Deficiency Syndrome 1	*SLC6A8*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 208,000
Cerebral Creatine Deficiency Syndrome 2	*GAMT*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,100
Cerebrotendinous Xanthomatosis	*CYP27A1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,900
Charcot-Marie-Tooth Disease, Type 4D	*NDRG1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 730,000
Charcot-Marie-Tooth Disease, Type 5 / Arts Syndrome	*PRPS1*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 114,000
Charcot-Marie-Tooth Disease, X-Linked	*GJB1*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 11,000
Choreoacanthocytosis	*VPS13A*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 13,000
Choroideremia	*CHM*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 125,000
Chronic Granulomatous Disease (*CYBA*-Related)	*CYBA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,700
Chronic Granulomatous Disease (*CYBB*-Related)	*CYBB*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 294,000
Citrin Deficiency	*SLC25A13*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 12,000
Citrullinemia, Type 1	*ASS1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,500
Cohen Syndrome	*VPS13B*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,700
Combined Malonic and Methylmalonic Aciduria	*ACSF3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,400
Combined Oxidative Phosphorylation Deficiency 1	*GFM1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 13,000
Combined Oxidative Phosphorylation Deficiency 3	*TSFM*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 27,000
Combined Pituitary Hormone Deficiency 2	*PROP1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,800
Combined Pituitary Hormone Deficiency 3	*LHX3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 140,000
Combined SAP Deficiency	*PSAP*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 44,000
Congenital Adrenal Hyperplasia due to 17-Alpha-Hydroxylase Deficiency	*CYP17A1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,800

Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency	*CYP21A2*	AR	Reduced Risk	*CYP21A2* copy number: 2 *CYP21A2* sequencing: Negative **Personalized Residual Risk (Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency (Non-Classic)):** 1 in 120 **Personalized Residual Risk (Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency (Classic)):** 1 in 780
Congenital Amegakaryocytic Thrombocytopenia	*MPL*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,100
Congenital Disorder of Glycosylation, Type Ia	*PMM2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 540
Congenital Disorder of Glycosylation, Type Ib	*MPI*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,600
Congenital Disorder of Glycosylation, Type Ic	*ALG6*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,100
Congenital Insensitivity to Pain with Anhidrosis	*NTRK1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,600
Congenital Myasthenic Syndrome (*CHRNE*-Related)	*CHRNE*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,100
Congenital Myasthenic Syndrome (*RAPSN*-Related)	*RAPSN*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,900
Congenital Neutropenia (*HAX1*-Related)	*HAX1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 82,000
Congenital Neutropenia (*VPS45*-Related)	*VPS45*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 43,000
Corneal Dystrophy and Perceptive Deafness	*SLC4A11*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,400
Corticosterone Methyloxidase Deficiency	*CYP11B2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,500
Cystic Fibrosis	*CFTR*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 440
Cystinosis	*CTNS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 7,700
D-Bifunctional Protein Deficiency	*HSD17B4*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,000
Deafness, Autosomal Recessive 77	*LOXHD1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,700
Duchenne Muscular Dystrophy / Becker Muscular Dystrophy	*DMD*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 10,000
Dyskeratosis Congenita (*RTEL1*-Related)	*RTEL1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 9,800
Dystrophic Epidermolysis Bullosa	*COL7A1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 900
Ehlers-Danlos Syndrome, Type VIIC	*ADAMTS2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 16,000
Ellis-van Creveld Syndrome (*EVC*-Related)	*EVC*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,200
Emery-Dreifuss Myopathy 1	*EMD*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 833,000
Enhanced S-Cone Syndrome	*NR2E3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,600
Ethylmalonic Encephalopathy	*ETHE1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,400
Fabry Disease	*GLA*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 7,700
Factor IX Deficiency	*F9*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 5,100
Factor XI Deficiency	*F11*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 730
Familial Autosomal Recessive Hypercholesterolemia	*LDLRAP1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 136,000
Familial Dysautonomia	*IKBKAP*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,400
Familial Hypercholesterolemia	*LDLR*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 280
Familial Hyperinsulinism (*ABCC8*-Related)	*ABCC8*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 450
Familial Hyperinsulinism (*KCNJ11*-Related)	*KCNJ11*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,300
Familial Mediterranean Fever	*MEFV*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 720
Fanconi Anemia, Group A	*FANCA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,100
Fanconi Anemia, Group C	*FANCC*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 8,100
Fanconi Anemia, Group G	*FANCG*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 12,000
Fragile X Syndrome	*FMR1*	XL	Reduced Risk	*FMR1* CGG repeat sizes: Not Performed *FMR1* Sequencing: Negative Fragile X CGG triplet repeat expansion testing was not performed at this time, as the patient has either been previously tested or is a male. **Personalized Residual Risk:** 1 in 8,300
Fumarase Deficiency	*FH*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,500
Galactokinase Deficiency	*GALK1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,700

sema4

Carrier screening report
Ian Kramer
Date of Birth: 02/19/1993
Sema4 ID: 22241019

Galactosemia	*GALT*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,200
Gaucher Disease	*GBA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 280
Gitelman Syndrome	*SLC12A3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 290
Glutaric Acidemia, Type I	*GCDH*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,800
Glutaric Acidemia, Type IIa	*ETFA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,700
Glutaric Acidemia, Type IIc	*ETFDH*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,700
Glycine Encephalopathy (*AMT*-Related)	*AMT*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,300
Glycine Encephalopathy (*GLDC*-Related)	*GLDC*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 760
Glycogen Storage Disease, Type Ia	*G6PC*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,300
Glycogen Storage Disease, Type Ib	*SLC37A4*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 7,300
Glycogen Storage Disease, Type II	*GAA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 520
Glycogen Storage Disease, Type IIi	*AGL*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,700
Glycogen Storage Disease, Type IV / Adult Polyglucosan Body Disease	*GBE1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,400
Glycogen Storage Disease, Type V	*PYGM*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 420
Glycogen Storage Disease, Type VII	*PFKM*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,300
GRACILE Syndrome and Other *BCS1L*-Related Disorders	*BCS1L*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,900
Hemochromatosis, Type 2A	*HFE2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,400
Hemochromatosis, Type 3	*TFR2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 7,400
Hereditary Fructose Intolerance	*ALDOB*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,900
Hereditary Spastic Paraparesis 49	*TECPR2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 15,000
Hermansky-Pudlak Syndrome, Type 1	*HPS1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,500
Hermansky-Pudlak Syndrome, Type 3	*HPS3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 27,000
HMG-CoA Lyase Deficiency	*HMGCL*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,700
Holocarboxylase Synthetase Deficiency	*HLCS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,500
Homocystinuria (*CBS*-Related)	*CBS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,400
Homocystinuria due to *MTHFR* Deficiency	*MTHFR*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,300
Homocystinuria, cblE Type	*MTRR*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 9,600
Hydrolethalus Syndrome	*HYLS1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 32,000
Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome	*SLC25A15*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,700
Hypohidrotic Ectodermal Dysplasia 1	*EDA*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 22,000
Hypophosphatasia	*ALPL*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 790
Inclusion Body Myopathy 2	*GNE*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,000
Infantile Cerebral and Cerebellar Atrophy	*MED17*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 129,000
Isovaleric Acidemia	*IVD*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,000
Joubert Syndrome 2	*TMEM216*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 14,000
Joubert Syndrome 7 / Meckel Syndrome 5 / COACH Syndrome	*RPGRIP1L*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 9,000
Junctional Epidermolysis Bullosa (*LAMA3*-Related)	*LAMA3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 9,300
Junctional Epidermolysis Bullosa (*LAMB3*-Related)	*LAMB3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,900
Junctional Epidermolysis Bullosa (*LAMC2*-Related)	*LAMC2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 77,000
Krabbe Disease	*GALC*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 860
Lamellar Ichthyosis, Type 1	*TGM1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,500
Leber Congenital Amaurosis 10 and Other CEP290-Related Ciliopathies	*CEP290*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,100
Leber Congenital Amaurosis 13	*RDH12*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,800
Leber Congenital Amaurosis 2 / Retinitis Pigmentosa 20	*RPE65*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,700
Leber Congenital Amaurosis 5	*LCA5*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 14,000

sema4

Carrier screening report
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Sema4 ID: 22241019

Leber Congenital Amaurosis 8 / Retinitis Pigmentosa 12 / Pigmented Paravenous Chorioretinal Atrophy	*CRB1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 990
Leigh Syndrome, French-Canadian Type	*LRPPRC*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 14,000
Lethal Congenital Contracture Syndrome 1 / Lethal Arthrogryposis with Anterior Horn Cell Disease	*GLE1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,900
Leukoencephalopathy with Vanishing White Matter	*EIF2B5*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,300
Limb-Girdle Muscular Dystrophy, Type 2A	*CAPN3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 960
Limb-Girdle Muscular Dystrophy, Type 2B	*DYSF*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,100
Limb-Girdle Muscular Dystrophy, Type 2C	*SGCG*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,900
Limb-Girdle Muscular Dystrophy, Type 2D	*SGCA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,500
Limb-Girdle Muscular Dystrophy, Type 2E	*SGCB*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 31,000
Limb-Girdle Muscular Dystrophy, Type 2I	*FKRP*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,400
Lipoamide Dehydrogenase Deficiency	*DLD*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,900
Lipoid Adrenal Hyperplasia	*STAR*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,600
Lipoprotein Lipase Deficiency	*LPL*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,600
Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	*HADHA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,900
Lysinuric Protein Intolerance	*SLC7A7*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,000
Maple Syrup Urine Disease, Type 1a	*BCKDHA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,100
Maple Syrup Urine Disease, Type 1b	*BCKDHB*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,100
Meckel Syndrome 1 / Bardet-Biedl Syndrome 13	*MKS1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,700
Medium Chain Acyl-CoA Dehydrogenase Deficiency	*ACADM*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,800
Megalencephalic Leukoencephalopathy with Subcortical Cysts	*MLC1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,300
Menkes Disease	*ATP7A*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 172,000
Metachromatic Leukodystrophy	*ARSA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,000
Methylmalonic Acidemia (*MMAA*-Related)	*MMAA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 15,000
Methylmalonic Acidemia (*MMAB*-Related)	*MMAB*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,700
Methylmalonic Acidemia (*MUT*-Related)	*MUT*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,300
Methylmalonic Aciduria and Homocystinuria, Cobalamin C Type	*MMACHC*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,800
Methylmalonic Aciduria and Homocystinuria, Cobalamin D Type	*MMADHC*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 219,000
Microphthalmia / Anophthalmia	*VSX2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 40,000
Mitochondrial Complex I Deficiency (*ACAD9*-Related)	*ACAD9*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,800
Mitochondrial Complex I Deficiency (*NDUFAF5*-Related)	*NDUFAF5*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 49,000
Mitochondrial Complex I Deficiency (*NDUFS6*-Related)	*NDUFS6*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 353,000
Mitochondrial DNA Depletion Syndrome 6 / Navajo Neurohepatopathy	*MPV17*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,400
Mitochondrial Myopathy and Sideroblastic Anemia 1	*PUS1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 333,000
Mucolipidosis II / IIIA	*GNPTAB*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,100
Mucolipidosis III Gamma	*GNPTG*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 51,000
Mucolipidosis IV	*MCOLN1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 9,400
Mucopolysaccharidosis Type I	*IDUA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,300
Mucopolysaccharidosis Type II	*IDS*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 76,000
Mucopolysaccharidosis Type IIIA	*SGSH*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,900
Mucopolysaccharidosis Type IIIB	*NAGLU*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 950
Mucopolysaccharidosis Type IIIC	*HGSNAT*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,200
Mucopolysaccharidosis Type IIID	*GNS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 137,000

sema4

Carrier screening report
Ian Kramer
Date of Birth: 02/19/1993
Sema4 ID: 22241019

Mucopolysaccharidosis Type IVb / GM1 Gangliosidosis	*GLB1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,700
Mucopolysaccharidosis type IX	*HYAL1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 149,000
Mucopolysaccharidosis type VI	*ARSB*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,300
Multiple Sulfatase Deficiency	*SUMF1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 30,000
Muscle-Eye-Brain Disease and Other *POMGNT1*-Related Congenital Muscular Dystrophy-Dystroglycanopathies	*POMGNT1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,200
Myoneurogastrointestinal Encephalopathy	*TYMP*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,100
Myotubular Myopathy 1	*MTM1*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 192,000
N-Acetylglutamate Synthase Deficiency	*NAGS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,200
Nemaline Myopathy 2	*NEB*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 660
Nephrogenic Diabetes Insipidus, Type II	*AQP2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,400
Nephrotic Syndrome (*NPHS1*-Related) / Congenital Finnish Nephrosis	*NPHS1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 920
Nephrotic Syndrome (*NPHS2*-Related) / Steroid-Resistant Nephrotic Syndrome	*NPHS2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 780
Neuronal Ceroid-Lipofuscinosis (*CLN3*-Related)	*CLN3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 9,200
Neuronal Ceroid-Lipofuscinosis (*CLN5*-Related)	*CLN5*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,300
Neuronal Ceroid-Lipofuscinosis (*CLN6*-Related)	*CLN6*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 8,600
Neuronal Ceroid-Lipofuscinosis (*CLN8*-Related)	*CLN8*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,100
Neuronal Ceroid-Lipofuscinosis (*MFSD8*-Related)	*MFSD8*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,200
Neuronal Ceroid-Lipofuscinosis (*PPT1*-Related)	*PPT1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 7,500
Neuronal Ceroid-Lipofuscinosis (*TPP1*-Related)	*TPP1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,300
Niemann-Pick Disease (*SMPD1*-Related)	*SMPD1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,800
Niemann-Pick Disease, Type C (*NPC1*-Related)	*NPC1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 690
Niemann-Pick Disease, Type C (*NPC2*-Related)	*NPC2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,600
Nijmegen Breakage Syndrome	*NBN*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 14,000
Non-Syndromic Hearing Loss (*GJB2*-Related)	*GJB2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 210
Omenn Syndrome (*RAG2*-Related)	*RAG2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 17,000
Omenn Syndrome / Severe Combined Immunodeficiency, Athabaskan-Type	*DCLRE1C*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,500
Ornithine Aminotransferase Deficiency	*OAT*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,400
Ornithine Transcarbamylase Deficiency	*OTC*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 103,000
Osteopetrosis 1	*TCIRG1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,700
Pendred Syndrome	*SLC26A4*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 390
Phenylalanine Hydroxylase Deficiency	*PAH*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 340
Polycystic Kidney Disease, Autosomal Recessive	*PKHD1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 450
Polyglandular Autoimmune Syndrome, Type 1	*AIRE*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,000
Pontocerebellar Hypoplasia, Type 1A	*VRK1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 25,000
Pontocerebellar Hypoplasia, Type 6	*RARS2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 8,600
Primary Carnitine Deficiency	*SLC22A5*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,500
Primary Ciliary Dyskinesia (*DNAH5*-Related)	*DNAH5*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,500
Primary Ciliary Dyskinesia (*DNAI1*-Related)	*DNAI1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,000
Primary Ciliary Dyskinesia (*DNAI2*-Related)	*DNAI2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 8,000
Primary Hyperoxaluria, Type 1	*AGXT*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,900
Primary Hyperoxaluria, Type 2	*GRHPR*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,500
Primary Hyperoxaluria, Type 3	*HOGA1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,400
Progressive Cerebello-Cerebral Atrophy	*SEPSECS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,400
Progressive Familial Intrahepatic Cholestasis, Type 2	*ABCB11*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 950
Propionic Acidemia (*PCCA*-Related)	*PCCA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,600

Propionic Acidemia (*PCCB*-Related)	*PCCB*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,100
Pycnodysostosis	*CTSK*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,100
Pyruvate Dehydrogenase E1-Alpha Deficiency	*PDHA1*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 139,000
Pyruvate Dehydrogenase E1-Beta Deficiency	*PDHB*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,000
Renal Tubular Acidosis and Deafness	*ATP6V1B1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,600
Retinitis Pigmentosa 25	*EYS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,800
Retinitis Pigmentosa 26	*CERKL*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,000
Retinitis Pigmentosa 28	*FAM161A*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 24,000
Retinitis Pigmentosa 59	*DHDDS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 9,900
Rhizomelic Chondrodysplasia Punctata, Type 1	*PEX7*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 10,000
Rhizomelic Chondrodysplasia Punctata, Type 3	*AGPS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 620,000
Roberts Syndrome	*ESCO2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 63,000
Salla Disease	*SLC17A5*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 8,400
Sandhoff Disease	*HEXB*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,800
Schimke Immunoosseous Dysplasia	*SMARCAL1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,800
Segawa Syndrome	*TH*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,100
Sjogren-Larsson Syndrome	*ALDH3A2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,500
Smith-Lemli-Opitz Syndrome	*DHCR7*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 750
Spinal Muscular Atrophy	*SMN1*	AR	Reduced Risk	*SMN1* copy number: 2 *SMN2* copy number: 1 c.*3+80T>G: Negative *SMN1* Sequencing: Negative **Personalized Residual Risk:** 1 in 1,107
Spondylothoracic Dysostosis	*MESP2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 225,000
Steel Syndrome	*COL27A1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 93,000
Stuve-Wiedemann Syndrome	*LIFR*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,000
Sulfate Transporter-Related Osteochondrodysplasia	*SLC26A2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,800
Tay-Sachs Disease	*HEXA*	AR	Reduced Risk	Tay-Sachs disease enzyme: Non-carrier White blood cells: Non-carrier • Hex A%: 67.8% (Non-carrier: 55.0 - 72.0%, Carrier: <50%) • Total hexosaminidase activity: 1933 nmol/hr/mg Plasma: Non-carrier • Hex A%: 65.1 (Non-carrier: 58.0 - 72.0%, Carrier: <54%) • Total hexosaminidase activity: 531 nmol/hr/ml *HEXA* Sequencing: Negative **Personalized Residual Risk:** 1 in 1,400
Tyrosinemia, Type I	*FAH*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,900
Usher Syndrome, Type IB	*MYO7A*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,000
Usher Syndrome, Type IC	*USH1C*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,600
Usher Syndrome, Type ID	*CDH23*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,400
Usher Syndrome, Type IF	*PCDH15*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,800
Usher Syndrome, Type IIA	*USH2A*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 290
Usher Syndrome, Type III	*CLRN1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,300
Very Long Chain Acyl-CoA Dehydrogenase Deficiency	*ACADVL*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 920
Walker-Warburg Syndrome and Other *FKTN*-Related Dystrophies	*FKTN*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,200
Wilson Disease	*ATP7B*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 350

Wolman Disease / Cholesteryl Ester Storage Disease	*LIPA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,200
X-Linked Juvenile Retinoschisis	*RS1*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 40,000
X-Linked Severe Combined Immunodeficiency	*IL2RG*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 250,000
Zellweger Syndrome Spectrum (*PEX10*-Related)	*PEX10*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,300
Zellweger Syndrome Spectrum (*PEX1*-Related)	*PEX1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,000
Zellweger Syndrome Spectrum (*PEX2*-Related)	*PEX2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 9,700
Zellweger Syndrome Spectrum (*PEX6*-Related)	*PEX6*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 910

AR=Autosomal recessive; XL=X-linked

Test methods and comments

Genomic DNA isolated from this patient was analyzed by one or more of the following methodologies, as applicable:

Fragile X CGG Repeat Analysis (Analytical Detection Rate >99%)

PCR amplification using Asuragen, Inc. AmplideX® *FMR1* PCR reagents followed by capillary electrophoresis for allele sizing was performed. Samples positive for *FMR1* premutations and full mutations greater than 90 CGG repeats in length were further analyzed by Southern blot analysis or methylation PCR to assess the size and methylation status of the *FMR1* CGG repeat. Additional testing to determine the status of AGG interruptions within the *FMR1* CGG repeat will be automatically performed for premutation alleles ranging from 55 to 90 repeats. These results, which may modify risk for expansion, will follow in a separate report.

Genotyping (Analytical Detection Rate >99%)

Multiplex PCR amplification and single-base pair probe extension analyses using the Agena Bioscience iPlex Pro chemistry on a MassARRAY® System were used to identify certain recurrent variants that are complex in nature or are present in low copy repeats. Rare sequence variants may interfere with assay performance.

Multiplex Ligation-Dependent Probe Amplification (MLPA) (Analytical Detection Rate >99%)

Conventional MLPA and/or digitalMLPA® probe sets and reagents from MRC-Holland were used for copy number variations (CNVs) analysis of specific targets versus known control samples. digitalMLPA® is a semi-quantitative technique, based on the well-established conventional MLPA method, followed by Illumina based sequencing to determine read number for amplicon quantification. False positive or negative results may occur due to rare sequence variants in target regions detected by conventional MLPA or digitalMLPA® probes. Analytical sensitivity and specificity of both the conventional MLPA method and the digitalMLPA® method are greater than 99%.

For alpha thalassemia, the copy numbers of the *HBA1* and *HBA2* genes were analyzed. Alpha-globin gene deletions, duplications, and the Constant Spring (CS) mutation are assessed. This test is expected to detect approximately 90% of all alpha-thalassemia mutations, varying by ethnicity. Carriers of alpha-thalassemia with three or more *HBA* copies on one chromosome, and one or no copies on the other chromosome, may not be precisely specified without phase analysis. With the exception of duplications, other benign alpha-globin gene polymorphisms will not be reported. Analyses of *HBA1* and *HBA2* are performed in association with long-range PCR of the coding regions followed by short-read sequencing.

For Duchenne muscular dystrophy, the copy numbers of all *DMD* exons were analyzed. Potentially pathogenic single exon deletions and duplications are confirmed by a second method. Analysis of *DMD* is performed in association with sequencing of the coding regions.

For congenital adrenal hyperplasia, the copy number of the *CYP21A2* gene was analyzed. This analysis can detect large deletions typically due to unequal meiotic crossing-over between *CYP21A2* and the pseudogene *CYP21A1P*. Classic 30-kb deletions make up approximately 20% of *CYP21A2* pathogenic alleles. This test may also identify certain point mutations in *CYP21A2* caused by gene conversion events between *CYP21A2* and *CYP21A1P*. Some carriers may not be identified by dosage sensitive methods as this testing cannot detect individuals with two copies (duplication) of the *CYP21A2* gene on one chromosome and loss of *CYP21A2* (deletion) on the other chromosome. Analysis of *CYP21A2* is performed in association with long-range PCR of the coding regions followed by short-read sequencing.

For spinal muscular atrophy (SMA), the copy numbers of the *SMN1* and *SMN2* genes were analyzed. The individual dosage of exons 7 and 8 as well as the combined dosage of exons 1, 4, 6 and 8 of *SMN1* and *SMN2* were assessed. Copy number gains and losses can be detected. Depending on ethnicity, 6 - 29 % of carriers will not be identified by dosage sensitive methods as this testing cannot distinguish individuals with two copies (duplication) of the *SMN1* gene on one chromosome and loss of *SMN1* (deletion) on the other chromosome (silent 2+0 carrier) or identify intragenic mutation in *SMN1*. Please also note that 2% of individuals diagnosed with SMA have a causative *SMN1* variant that occurred

de novo, therefore cannot be picked up by carrier screening in the parents. Analysis of *SMN1* is performed in association with short-read sequencing of exons 2a-7, followed by confirmation using long-range PCR (described below).

In individuals with two copies of *SMN1* with Ashkenazi Jewish, East Asian, African American, Native American or Caucasian ancestry, the presence or absence of c.*3+80T>G significantly increases or decreases, respectively, the likelihood of being a silent 2+0 silent carrier.

MLPA for Gaucher disease *(GBA)*, cystic fibrosis *(CFTR)*, and non-syndromic hearing loss *(GJB2/GJB6)* will only be performed if indicated for confirmation of detected CNVs. If *GBA* analysis was performed, the copy numbers of exons 1, 3, 4, and 6 - 10 of the GBA gene (of 11 exons total) were analyzed. If *CFTR* analysis was performed, the copy numbers of all 27 *CFTR* exons were analyzed. If *GJB2/GJB6* analysis was performed, the copy number of the two *GJB2* exons were analyzed, as well as the presence or absence of the two upstream deletions of the *GJB2* regulatory region, del(*GJB6*-D13S1830) and del(*GJB6*-D13S1854).

Next Generation Sequencing (NGS) (Analytical Detection Rate >95%)

NGS was performed on a panel of genes for the purpose of identifying pathogenic or likely pathogenic variants.

Agilent SureSelect^{TM}XT Low Input technology was used with a custom capture library to target the exonic regions and intron/exon splice junctions of the relevant genes, as well as a number of UTR, intronic or promoter regions that contain previously reported mutations. Libraries were pooled and sequenced on the Illumina NovaSeq 6000 platform, using paired-end 100 bp reads. The sequencing data was analyzed using a custom bioinformatics algorithm designed and validated in house.

The coding exons and splice junctions of the known protein-coding RefSeq genes were assessed for the average depth of coverage (minimum of 20X) and data quality threshold values. Most exons not meeting a minimum of >20X read depth across the exon are further analyzed by Sanger sequencing. Please note that several genomic regions present difficulties in mapping or obtaining read depth >20X. These regions, which are described below, will not be reflexed to Sanger sequencing if the mapping quality or coverage is poor. Any variants identified during testing in these regions are confirmed by a second method and reported if determined to be pathogenic or likely pathogenic. However, as there is a possibility of false negative results within these regions, detection rates and residual risks for these genes have been calculated with the presumption that variants in these exons will not be detected, unless included in the MassARRAY® genotyping platform.

Exceptions: *ABCD1* (NM_000033.3) exons 8 and 9; *ACADSB* (NM_ 001609.3) chr10:124,810,695-124,810,707 (partial exon 9); *ADA* (NM_000022.2) exon 1; *ADAMTS2* (NM_014244.4) exon 1; *AGPS* (NM_003659.3) chr2:178,257,512-178,257,649 (partial exon 1); *ALDH7A1* (NM_001182.4) chr5:125,911,150-125,911,163 (partial exon 7) and chr5:125,896,807-125,896,821 (partial exon 10); *ALMS1* (NM_015120.4) chr2:73,612,990-73,613,041 (partial exon 1); *APOPT1* (NM_ 032374.4) chr14:104,040,437-104,040,455 (partial exon 3); *CDAN1* (NM_138477.2) exon 2; *CEP152* (NM_014985.3) chr15:49,061,146-49,061,165 (partial exon 14) and exon 22; *CEP290* (NM_025114.3) exon 5, exon 7, chr12:88,519,017-88,519,039 (partial exon 13), chr12:88,514,049-88,514,058 (partial exon 15), chr12:88,502,837-88,502,841 (partial exon 23), chr12:88,481,551-88,481,589 (partial exon 32), chr12:88,471,605-88,471,700 (partial exon 40); *CFTR* (NM_000492.3) exon 10; *COL4A4* (NM_000092.4) chr2:227,942,604-227,942,619 (partial exon 25); *COX10* (NM_001303.3) exon 6; *CYP11B1* (NM_000497.3) exons 3-7; *CYP11B2* (NM_000498.3) exons 3-7; *DNAI2* (NM_023036.4) chr17:72,308,136-72,308,147 (partial exon 12); *DOK7* (NM_173660.4) chr4:3,465,131-3,465,161 (partial exon 1) and exon 2; *DUOX2* (NM_014080.4) exons 6-8; *EIF2AK3* (NM_004836.5 exon 8; *EVC* (NM_153717.2) exon 1; *F5*(NM_000130.4) chr1:169,551,662-169,551,679 (partial exon 2); *FH* (NM_000143.3) exon 1; *GAMT* (NM_000156.5 exon 1; *GLDC*(NM_000170.2) exon 1; *GNPTAB* (NM_024312.4) chr17:4,837,000-4,837,400 (partial exon 2); *GNPTG* (NM_032520.4) exon 1; *GHR* (NM_000163.4) exon 3; *GYS2* (NM_021957.3) chr12:21,699,370-21,699,409 (partial exon 12); *HGSNAT* (NM_152419.2) exon 1; *IDS* (NM_000202.6 exon 3; *ITGB4* (NM_000213.4) chr17:73,749,976-73,750,060 (partial exon 33); *JAK3* (NM_000215.3) chr19:17,950,462-17,950,483 (partial exon 10); *LIFR* (NM_002310.5 exon 19; *LMBRD1* (NM_018368.3) chr6:70,459,226-70,459,257 (partial exon 5), chr6:70,447,828-70,447,836 (partial exon 7) and exon 12; *LYST* (NM_000081.3) chr1:235,944,158-235,944,176 (partial exon 16) and chr1:235,875,350-235,875,362 (partial exon 43); *MLYCD* (NM_012213.2) chr16:83,933,242-83,933,282 (partial exon 1); *MTR* (NM_000254.2) chr1 237,024,418-237,024,439 (partial exon 20) and chr1:237,038,019-237,038,029 (partial exon 24); *NBEAL2* (NM_015175.2) chr3 47,021,385-47,021,407 (partial exon 1); *NEB* (NM_001271208.1 exons 82-105; *NPC1* (NM_000271.4) chr18:21,123,519-21,123,538 (partial exon 14); *NPHP1* (NM_000272.3)chr2:110,937,251-110,937,263 (partial exon 3); *OCRL* (NM_000276.3) chrX:128,674,450-128,674,460 (partial exon 1); *PHKB* (NM_000293.2) exon 1 and chr16:47,732,498-47,732,504 (partial exon 30); *PIGN* (NM_176787.4) chr18:59,815,547-59,815,576 (partial exon 8); *PIP5K1C* (NM_012398.2) exon 1 and chr19:3637602-3637616 (partial exon 17); *POU1F1* (NM_000306.3) exon 5; *PTPRC* (NM_002838.4) exons 11 and 23; *PUS1* (NM_025215.5 chr12:132,414,446-132,414,532 (partial exon 2); *RPGRIP1L* (NM_015272.2) exon 23; *SGSH* (NM_000199.3) chr17:78,194,022-78,194,072 (partial exon 1); *SLC6A8* (NM_005629.3) exons 3 and 4; *ST3GAL5* (NM_003896.3) exon 1; *SURF1* (NM_003172.3) chr9:136,223,269-136,223,307 (partial exon 1); *TRPM6* (NM_017662.4) chr9:77,362,800-77,362,811 (partial exon 31); *TSEN54* (NM_207346.2) exon 1; *TYR* (NM_000372.4) exon 5; *VWF* (NM_000552.3) exons 24-26, chr12:6,125,675-6,125,684 (partial exon 30), chr12:6,121,244-6,121,265 (partial exon 33), and exon 34.

This test will detect variants within the exons and the intron-exon boundaries of the target regions. Variants outside these regions may not be detected, including, but not limited to, UTRs, promoters, and deep intronic areas, or regions that fall into the Exceptions mentioned above. This technology may not detect all small insertion/deletions and is not diagnostic for repeat expansions and structural genomic variation. In addition, a mutation(s) in a gene not included on the panel could be present in this patient.

Variant interpretation and classification was performed based on the American College of Medical Genetics Standards and Guidelines for the Interpretation of Sequence Variants (Richards et al. 2015). All potentially pathogenic variants may be confirmed by either a specific genotyping assay or Sanger sequencing, if indicated. Any benign variants, likely benign variants or variants of uncertain significance identified during this analysis will not be reported.

Next Generation Sequencing for *SMN1*

Exonic regions and intron/exon splice junctions of *SMN1* and *SMN2* were captured, sequenced, and analyzed as described above. Any variants located within exons 2a-7 and classified as pathogenic or likely pathogenic were confirmed to be in either *SMN1* or *SMN2* using gene-specific long-range PCR analysis followed by Sanger sequencing. Variants located in exon 1 cannot be accurately assigned to either *SMN1* or *SMN2* using our current methodology, and so these variants are not reported.

Copy Number Variant (CNV) Analysis (Analytical Detection Rate >98% for CNVs of 3 exons and larger, >90% for CNVs of 2 exons)

Large duplications and deletions were called from the relative read depths on an exon-by-exon basis using a custom exome hidden Markov model (XHMM) algorithm. Deletions or duplications determined to be pathogenic or likely pathogenic were confirmed by either a custom arrayCGH platform, quantitative PCR, or MLPA (depending on CNV size and gene content). While this algorithm is designed to pick up deletions and duplications of 2 or more exons in length, potentially pathogenic single-exon CNVs will be confirmed and reported, if detected. Deletions and duplications near the lower limit of detection may not be detected due to run variability. Genomic regions with high homology or highly repetitive sequences are excluded from this analysis.

Exon Array Comparative Genomic Hybridization (aCGH) (Confirmation method) (Accuracy >99%)

The customized oligonucleotide microarray (Oxford Gene Technology) is a highly-targeted exon-focused array capable of detecting medically relevant microdeletions and microduplications at a much higher resolution than traditional aCGH methods. Each array matrix has approximately 1,000,000 60-mer oligonucleotide probes that cover the entire genome. This platform is designed based on human genome NCBI Build 37 (hg19) and the CGH probes are enriched to target the exonic regions of the genes in this panel.

Quantitative PCR (Confirmation method) (Accuracy >99%)

The relative quantification PCR is utilized on a Roche SYBR Green reagents on a LightCycler® 480 System, which relates the PCR signal of the target region in one group to another. To test for genomic imbalances, both sample DNA and reference DNA is amplified with primer/probe sets that specific to the target region and a control region with known genomic copy number. Relative genomic copy numbers are calculated based on the standard ΔΔCt formula.

Long-Range PCR (Analytical Detection Rate >99%)

Long-range PCR was performed to generate locus-specific amplicons for *CYP21A2*, *HBA1* and *HBA2* and *GBA*. The PCR products were then prepared for short-read NGS sequencing and sequenced. Sequenced reads were mapped back to the original genomic locus and run through the bioinformatics pipeline. If indicated, copy number from MLPA was correlated with the sequencing output to analyze the results. Please note that in rare cases, allele drop-out may occur, which has the potential to lead to false negative results. For *CYP21A2*, a certain percentage of healthy individuals carry a duplication of the *CYP21A2* gene, which has no clinical consequences. In cases where multiple copies of *CYP21A2* are located on the same chromosome in tandem, only the last copy will be amplified and assessed for potentially pathogenic variants, due to size limitations of the PCR reaction. However, because these alleles contain at least two copies of the *CYP21A2* gene in tandem, it is expected that this patient has at least one functional gene in the tandem allele and this patient is therefore less likely to be a carrier. A *CYP21A1P/CYP21A2* hybrid gene detected only by MLPA but not by long-range PCR will not be reported when the long-range PCR indicates the presence of two full *CYP21A2* gene copies (one on each chromosome), as the additional hybrid gene is nonfunctional. Classic 30-kb deletions are identified by MLPA and are also identified by the presence of multiple common pathogenic *CYP21A2* variants by long-range PCR. Since multiple pseudogene-derived variants are detected in all cases with the classic 30kb deletion, we cannot rule out the possibility that some variant(s) detected could be present in trans with the chimeric *CYP21A1P/CYP21A2* gene created by the 30kb deletion. When an individual carries both a duplication allele and a pathogenic variant, or multiple pathogenic variants, the current analysis may not be able to determine the phase (cis/trans configuration) of the *CYP21A2* alleles identified. Family studies may be required in certain scenarios where phasing is required to determine the carrier status.

Residual Risk Calculations

Carrier frequencies and detection rates for each ethnicity were calculated through the combination of internal curations of >30,000 variants and genomic frequency data from >138,000 individuals across seven ethnic groups in the gnomAD database. Additional variants in HGMD and novel deleterious variants were also incorporated into the calculation. Residual risk values are calculated using a Bayesian analysis combining the a *priori* risk of being a pathogenic mutation carrier (carrier frequency) and the detection rate. They are provided only as a guide for assessing approximate risk given a negative result, and values will vary based on the exact ethnic background of an individual. This report does

not represent medical advice but should be interpreted by a genetic counselor, medical geneticist or physician skilled in genetic result interpretation and the relevant medical literature.

Personalized Residual Risk Calculations

Agilent SureSelect^{TM}XT Low-Input technology was utilized in order to create whole-genome libraries for each patient sample. Libraries were then pooled and sequenced on the Illumina NovaSeq platform. Each sequencing lane was multiplexed to achieve 0.4-2x genome coverage, using paired-end 100 bp reads. The sequencing data underwent ancestral analysis using a customized, licensed bioinformatics algorithm that was validated in house. Identified sub-ethnic groupings were binned into one of 7 continental-level groups (African, East Asian, South Asian, Non-Finnish European, Finnish, Native American, and Ashkenazi Jewish) or, for those ethnicities that matched poorly to the continental-level groups, an 8th "unassigned" group, which were then used to select residual risk values for each gene. For individuals belonging to multiple high-level ethnic groupings, a weighting strategy was used to select the most appropriate residual risk. For genes that had insufficient data to calculate ethnic-specific residual risk values, or for sub-ethnic groupings that fell into the "unassigned" group, a "worldwide" residual risk was used. This "worldwide" residual risk was calculated using data from all available continental-level groups.

Several genes have multiple residual risks associated to reflect the likelihood of the tested individual being a carrier for different diseases that are attributed to non-overlapping pathogenic variants in that gene. When calculating the couples' combined reproductive risk, the highest residual risk for each patient was selected.

Sanger Sequencing (Confirmation method) (Accuracy >99%)

Sanger sequencing, as indicated, was performed using BigDye Terminator chemistry with the ABI 3730 DNA analyzer with target specific amplicons. It also may be used to supplement specific guaranteed target regions that fail NGS sequencing due to poor quality or low depth of coverage (<20 reads) or as a confirmatory method for NGS positive results. False negative results may occur if rare variants interfere with amplification or annealing.

Tay-Sachs Disease (TSD) Enzyme Analysis (Analytical Detection Rate ≥98%)

Hexosaminidase activity and Hex A% activity were measured by a standard heat-inactivation, fluorometric method using artificial 4-MU-β-N-acetyl glucosaminide (4-MUG) substrate. This assay is highly sensitive and accurate in detecting Tay-Sachs carriers and individuals affected with TSD. Normal ranges of Hex A% activity are 55.0-72.0 for white blood cells and 58.0-72.0 for plasma. It is estimated that less than 0.5% of Tay-Sachs carriers have non-carrier levels of percent Hex A activity, and therefore may not be identified by this assay. In addition, this assay may detect individuals that are carriers of or are affected with Sandhoff disease. False positive results may occur if benign variants, such as pseudodeficiency alleles, interfere with the enzymatic assay. False negative results may occur if both *HEXA* and *HEXB* pathogenic or pseudodeficiency variants are present in the same individual.

Please note that it is not possible to perform Tay-Sachs disease enzyme analysis on saliva samples, buccal swabs, tissue samples, semen samples, or on samples received as extracted DNA.

This test was developed, and its performance characteristics determined by Sema4 Opco, Inc. It has not been cleared or approved by the US Food and Drug Administration. FDA does not require this test to go through premarket FDA review. This test is used for clinical purposes. It should not be regarded as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments (CLIA) as qualified to perform high complexity clinical laboratory testing. These analyses generally provide highly accurate information regarding the patient's carrier or affected status. Despite this high level of accuracy, it should be kept in mind that there are many potential sources of diagnostic error, including misidentification of samples, polymorphisms, or other rare genetic variants that interfere with analysis. Families should understand that rare diagnostic errors may occur for these reasons.

SELECTED REFERENCES

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Fragile X syndrome:

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Scott SA et al. Experience with carrier screening and prenatal diagnosis for sixteen Ashkenazi Jewish Genetic Diseases. *Hum. Mutat.* 2010 31:1-11.

Akler G et al. Towards a unified approach for comprehensive reproductive carrier screening in the Ashkenazi, Sephardi, and Mizrahi Jewish populations. *Mol Genet Genomic Med.* 2020 Feb 8(2):e1053.

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Flanigan KM et al. Mutational spectrum of *DMD* mutations in dystrophinopathy patients: application of modern diagnostic techniques to a large cohort. *Hum Mutat.* 2009 30:1657-66.

Variant Classification:

Richards S et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. *Genet Med.* 2015 May;17(5):405-24.

Riggs ER, Andersen EF, Cherry AM, et al. Technical standards for the interpretation and reporting of constitutional copy-number variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics (ACMG) and the Clinical Genome Resource (ClinGen) [published correction appears in Genet Med. 2021 Nov;23(11):2230]. Genet Med. 2020;22(2):245-257.

Additional disease-specific references available upon request.

rint date: 8/19/22 15:16 P A T I E N T R E P O R T Page 1
rinted by: **** FINAL ****

RESTWOOD MEDICAL CENTER LAB - CLINICAL MAIN
NE HOSPITAL DRIVE
UNTSVILLE AL 35801

ame: KRAMER IAN A Status: O/P / LOP Adm Date: 8/19/22
at#: 3010127 DOB: 2/19/93 Adm Phys: GENNARO KYLE
trt: 8/19/22 10:56 Age/Sex: 29 / M Ord Phys: GENNARO KYLE
rd#: R 100 MR#: 000883262 Fam Phys: CAMPBELL YARI P

pecial Instructions:
Reported: 8/19/22 15:16

est Name	Result	Flag	Reference Range	Units
ollected: 8/19/22 9:40 CBN Received: 8/19/22 10:56 KB1 Verified: 8/19/22 15:16 HL7				
EMEN ANAL COMP				
ABSTINENCE	4		2 - 7	days
VOLUME	3.0		>1.9	mL
PH	8.5		>7.1	
LIQUEFACTION	TNP		0 - 60	min
SPERM COUNT	19.00	L	>19.90	mill/mL
WBC	322		0 - 1000	per uL
VIABILITY	TNP		>49	%
MOTILITY	42	L	>49	%
MORPHOLOGY	22		>15	%

OMMENTS:
8/19/22 15:16 SEMCWDIF TNP = Test Not Performed. Any calculations using this test cannot be performed.
LIQUIFAC Specimen would not liquefy completely after 2 1/2 hours
VIABIL Unable to perform test due to liquefacation issues

IntrinsiQ Specialty Solutions

FAX COVER SHEET

To:	Alabama		
Company:	Alabama	Date:	11/15/2022 9:59 AM
Phone:		Fax:	12058747021
Pages:	2 (including cover page)		
From:	Ella Watts on behalf of	Fax:	1(417) 507-1786
Phone:	2568823605		
Practice:	Client - Urology Specialists - 35289		

Subject:

Message:

Referring Physician: Mann, Merry Lynn

Test Date: 11/29/2022

Patient: KRAMER, IAN
Patient DOB: 02/19/1993
Patient CLN: 400534F93
Sex: Male

Partner : JENNIS, ARIEL
Partner DOB: 04/16/1992
Partner CLN: CL-257AJ-1777666
Sex: Female

Medication (Prescription and Non-Prescription): ☐ Ca+ Channel Blocker ☐ Blood Pressure Medication ☐ Testosterone ☐ Proscar ☐ Marijuana ☐ Other Recreational Drugs ☑ N/A

Other Medication: Adderall

Medical Procedures: ☐ Vasectomy ☐ Vasectomy Repair ☐ Other Reproduction Tract Surgery ☐ Radiation Therapy ☐ Chemotherapy ☑ N/A

Recent Health: Have you been sick in the last three months? ☐ Yes ☑ No ☐ Fever?

Recent Health Comments

Date Of Collection	11/29/2022	Collection Time	10:07AM	Donor Type	Partner		
Received at lab by		Time Sample Received	10:11AM	Location of Collection	In Office	Container type	Sterile Cup/Sen
Days Abstinence	5 days			Analyzed and Processed by		Time of analysis	10:41AM
Method of Collection	Masturbation			Complete Ejaculate?	Yes	Spillage Percent (%)	0%
Type S/A	☑ Macroscopic	Analysis Tempature	☐ RT	Liquefaction (>2 cm)	**Complete by 60**	Viscosity	☑ < 2 cm ☐ > 2 cm
Agglutination– None	None	Leuco Screen; <1 x 10^6/mL	(<1 x 10^6/mL)		Pentox; N/A		
Round Cells (10^6/mL); <4 x10^6/mL	0.9						

Morphology: ☑ Normal ☑ Tapered Heads ☑ Pyriform Heads ☑ Amorphous Heads ☑ Neck Defects ☑ Tail Defects ☐ Coiled Tails ☐ Double Head/Tails

Morphology (%); >4%	(>4%) **1**	Morphology Tech	Conville, Hanna	pH >7.2	(>=7.2)	8.5	
Sample Color	☐ White ☑ Grey ☐ Yellow ☐ Clear ☐ Brown ☐ Red						
Characteristics							
Volume	(<1.0mL) 3ml mL	Sperm Concentration	24 Mill/mL	Total sperm in ejaculate	72 Million		
Total Motility	59% %	Total motile	42.48 Million	Progression %; P>32%	(>=32%) **44%**		
Non-Progression %	(>=32%) **13%**	Non-Motile %	(>=32%) **43%**				

Comments: Joan Reed Np was the analyst. Spoke to patient and spouse, reviewed SA results in detail, informed morphology still pending.

IVF or IUI Indicated.

*WHO laboratory examination of human semen and sperm cervical mucus interaction, 5th edition, 2010.h

Requesting Physician: Dr Mann

Technician: Time Complete 11:45AM

Report Finalized by: Hannah Conville on 12/01/2022 09:07 AM

End of **Final** report

Signature:

Referring Physician: Mann, Merry Lynn

Test Date: 02/15/2023

Patient: KRAMER, IAN
Patient DOB: 02/19/1993
Patient CLN: 400534F93
Sex: Male

Partner : JENNIS, ARIEL
Partner DOB: 04/16/1992
Partner CLN: CL-257AJ-1777666
Sex: Female

Medication (Prescription and Non-Prescription) ☐ Ca+ Channel Blocker ☐ Blood Pressure Medication ☐ Testosterone ☐ Proscar ☐ Marijuana ☐ Other Recreational Drugs ☑ N/A

Other Medication Adderall

Medical Procedures ☐ Vasectomy ☐ Vasectomy Repair ☐ Other Reproduction Tract Surgery ☐ Radiation Therapy ☐ Chemotherapy ☑ N/A

Recent Health: Have you been sick in the last three months? ☐ Yes ☑ No ☐ Fever?

Recent Health Comments

Date Of Collection	02/15/2023	Collection Time	7:30AM	Donor Type	Partner		
Received at lab by	Goodman, Heat	Time Sample Received	7:43AM	Location of Collection	Other	Container type	Sterile Cup/Sen
Days Abstinence	3 days			Analyzed and Processed by		Time of analysis	8:00AM
Method of Collection	Masturbation			Complete Ejaculate?	No, 1st portion	Spillage Percent (%)	10%
Analysis Tempature	☑ RT	Liquefaction (>2 cm)	**Complete by 60**	Viscosity	☑ < 2 cm ☐ > 2 cm		
Agglutination– None	None	Characteristics					
Sample Color	☐ White ☑ Grey ☐ Yellow ☐ Clear ☐ Brown ☐ Red			Sperm Volume (mL)	1.5ml		
Sperm Concentration	13	Sperm Motility	72%	Total Motile Sperm Count	14.04		

Comments: Joan Reed Np was the analyst, Np reviewed IUI Sample data in detail with patient prior to insemination. Aware count on low end but IUI still appropriate to proceed. Patient verbalized understanding.

Volume	0.5ml mL	Sperm Concentration	9 Mill/mL	Total Motility	72% %
Total motile	3.24 Million				

*WHO laboratory examination of human semen and sperm cervical mucus interaction, 5th edition, 2010.h

Time Complete 8:35AM Technician

Secondary Witness: Mary Duram

Report Finalized by: Joan Reed on 02/16/2023 08:21 AM

End of **Final** report

Signature: _____

Referring Physician: Mann, Merry Lynn

Test Date: 04/11/2023

Patient: KRAMER, IAN
Patient DOB: 02/19/1993
Patient CLN: 400534F93
Sex: Male

Partner : JENNIS, ARIEL
Partner DOB: 04/16/1992
Partner CLN: CL-257AJ-1777666
Sex: Female

Medication (Prescription and Non-Prescription): ☐ Ca+ Channel Blocker ☐ Blood Pressure Medication ☐ Testosterone ☐ Proscar ☐ Marijuana ☐ Other Recreational Drugs ☑ N/A

Other Medication: Adderall 15 mg

Medical Procedures: ☐ Vasectomy ☐ Vasectomy Repair ☐ Other Reproduction Tract Surgery ☐ Radiation Therapy ☐ Chemotherapy ☑ N/A

Recent Health: Have you been sick in the last three months? ☐ Yes ☑ No ☐ Fever?

Recent Health Comments: Admits to being sick last IUI.

Date Of Collection	04/11/2023	Collection Time	11:42AM	Donor Type	Partner		
Received at lab by	Goodman, Heat	Time Sample Received	11:42AM	Location of Collection	In Office	Container type	Sterile Cup/Sen
Days Abstinence	6 days			Analyzed and Processed by		Time of analysis	11:52AM
Method of Collection	Masturbation			Complete Ejaculate?	No, 1st portion	Spillage Percent (%)	10
Analysis Tempature	☑ RT	Liquefaction (>2 cm)	**Complete by 60**	Viscosity	☑ < 2 cm ☐ > 2 cm		
Agglutination– None	None	Characteristics					
Sample Color	☐ White ☑ Grey ☐ Yellow ☐ Clear ☐ Brown ☐ Red			Sperm Volume (mL)	2		
Sperm Concentration	520	Sperm Motility	69	Total Motile Sperm Count	717.6		

Comments: Analyzed sample J.Reed CRNP/S.Callahan, RN. IUI sample reviewed with patient and spouse in clinic prior to insemination

Volume	0.5 mL	Sperm Concentration	93 Mill/mL	Total Motility	82 %
Total motile	38.13 Million				

*WHO laboratory examination of human semen and sperm cervical mucus interaction, 5th edition, 2010.h

Time Complete	12:30PM	Technician		Secondary Witness:	J.Reed, CRNP, S.Callal

Report Finalized by: Joan Reed on 04/11/2023 01:55 PM

End of **Final** report

Signature: _____

Referring Physician: Mann, Merry Lynn

Test Date: 07/20/2023

Patient: KRAMER, IAN
Patient DOB: 02/19/1993
Patient CLN: 400534F93
Sex: Male

Partner : JENNIS, ARIEL
Partner DOB: 04/16/1992
Partner CLN: CL-257AJ-1777666
Sex: Female

Medication (Prescription and Non-Prescription): ☐ Ca+ Channel Blocker ☐ Blood Pressure Medication ☐ Testosterone ☐ Proscar ☐ Marijuana ☐ Other Recreational Drugs ☑ N/A

Other Medication:

Medical Procedures: ☐ Vasectomy ☐ Vasectomy Repair ☐ Other Reproduction Tract Surgery ☐ Radiation Therapy ☐ Chemotherapy ☑ N/A

Recent Health: Have you been sick in the last three months? ☐ Yes ☑ No ☐ Fever?

Recent Health Comments: N/A

Date Of Collection	07/20/2023	Collection Time	11:35AM	Donor Type			
Received at lab by	Jones, Teri	Time Sample Received	11:42AM	Location of Collection	In Office	Container type	Sterile Cup/Sen
Days Abstinence	5 days			Analyzed and Processed by	Jones, Teri	Time of analysis	12:18PM
Method of Collection	Masturbation			Complete Ejaculate?	Yes	Spillage Percent (%)	N/A
Analysis Tempature	☑ RT	Liquefaction (>2 cm)	VTS	Viscosity	☐ < 2 cm ☑ > 2 cm		
Agglutination– None		Characteristics					
Sample Color	☐ White ☐ Grey ☐ Yellow ☐ Clear ☐ Brown ☐ Red			Sperm Volume (mL)	3.5		
Sperm Concentration	34.5	Sperm Motility	48	Total Motile Sperm Count	57.96		

Comments: VTS USED

Volume	0.5 mL	Sperm Concentration	49.5 Mill/mL	Total Motility	47 %
Total motile	11.63 Million				

*WHO laboratory examination of human semen and sperm cervical mucus interaction, 5th edition, 2010.h

Time Complete	12:47PM	Technician	Jones, Teri	Secondary Witness:	MH

Report Finalized by: Teri Jones on 07/20/2023 12:45 PM

End of **Final** report

Signature: _____

Referring Physician: Mann, Merry Lynn, MD

Test Date: 09/28/2023

Patient: KRAMER, IAN
Patient DOB: 02/19/1993
Patient CLN: 400534F93
Sex: Male

Partner : JENNIS, ARIEL
Partner DOB: 04/16/1992
Partner CLN: CL-257AJ-1777666
Sex: Female

Medication (Prescription and Non-Prescription): ☐ Ca+ Channel Blocker ☐ Blood Pressure Medication ☐ Testosterone ☐ Proscar ☐ Marijuana ☐ Other Recreational Drugs ☐ N/A

Other Medication: Adderall

Medical Procedures: ☐ Vasectomy ☐ Vasectomy Repair ☐ Other Reproduction Tract Surgery ☐ Radiation Therapy ☐ Chemotherapy ☑ N/A

Recent Health: Have you been sick in the last three months? ☐ Yes ☑ No ☐ Fever?

Recent Health Comments:

Date Of Collection	09/28/2023	Collection Time	8:18AM	Received at lab by	Cook, Maggie	Time Sample Received	8:20AM
Location of Collection	In Office	Container type	Sterile Cup/Sen	Days Abstinence	3 days		
Analyzed and Processed by	Sullivan, Leslie	Time of analysis	8:50AM	Method of Collection		Masturbation	
Complete Ejaculate?	Yes	Spillage Percent (%)	0	Analysis Tempature	☑ RT	Liquefaction (>2 cm)	**VTS**
Viscosity	☐ < 2 cm ☑ > 2 cm	Agglutination– None	None	Characteristics			
Sample Color	☐ White ☐ Grey ☑ Yellow ☐ Clear ☐ Brown ☐ Red			Volume (<1.0mL)	**3.4 mL**		
Sperm Concentration	24.0 Mill/mL	Total Motility	42 %	Total sperm in ejaculate	81.60 Million		
Total motile	34.27 Million	Progression	42	Non-Progression	2		
Non-Motile	56						

Comments:

*WHO laboratory examination of human semen and sperm cervical mucus interaction, 5th edition, 2010.h

Time Complete: Technician: Sullivan, Leslie Secondary Witness: LJ

Report Finalized by: Leslie Sullivan on 09/28/2023 11:53 AM

End of **Final** report

Signature: _____

Referring Physician: Mann, Merry Lynn, MD

Test Date: 11/15/2023

Patient: KRAMER, IAN
Patient DOB: 02/19/1993
Patient CLN: 400534F93
Sex: Male

Partner : JENNIS, ARIEL
Partner DOB: 04/16/1992
Partner CLN: CL-257AJ-1777666
Sex: Female

Medication (Prescription and Non-Prescription): ☐ Ca+ Channel Blocker ☐ Blood Pressure Medication ☐ Testosterone ☐ Proscar ☐ Marijuana ☐ Other Recreational Drugs ☑ N/A

Other Medication:

Medical Procedures: ☐ Vasectomy ☐ Vasectomy Repair ☐ Other Reproduction Tract Surgery ☐ Radiation Therapy ☐ Chemotherapy ☑ N/A

Recent Health: Have you been sick in the last three months? ☐ Yes ☑ No ☐ Fever?

Recent Health Comments:

Date Of Collection	11/15/2023	Collection Time	9:45AM	Received at lab by	Jones, Teri	Time Sample Received	9:46AM
Location of Collection	In Office	Container type	Sterile Cup/Sen	Days Abstinence	2 days		
Analyzed and Processed by	Sullivan, Leslie	Time of analysis	10:20AM	Method of Collection		Masturbation	
Complete Ejaculate?	Yes	Spillage Percent (%)	0	Analysis Tempature	☑ RT	Liquefaction (>2 cm)	**VTS**
Viscosity	☐ < 2 cm ☑ > 2 cm	Agglutination– None	None	Characteristics			
Sample Color	☐ White ☐ Grey ☑ Yellow ☐ Clear ☐ Brown ☐ Red	Volume (<1.0mL)	**3.0 mL**				

Sperm Concentration	25.5 Mill/mL	Total Motility	45 %	Total sperm in ejaculate	76.50 Million
Total motile	34.42 Million	Progression	40	Non-Progression	5
Non-Motile	55				

Comments:

*WHO laboratory examination of human semen and sperm cervical mucus interaction, 5th edition, 2010.h

Time Complete: | Technician: Sullivan, Leslie | Secondary Witness: LJ

Report Finalized by: Leslie Sullivan on 11/15/2023 11:30 AM

End of **Final** report

Signature: _____

Referring Physician: Mann, Merry Lynn, MD

Test Date: 12/13/2023

Patient: KRAMER, IAN
Patient DOB: 02/19/1993
Patient CLN: 400534F93
Sex: Male

Partner : JENNIS, ARIEL
Partner DOB: 04/16/1992
Partner CLN: CL-257AJ-1777666
Sex: Female

Medication (Prescription and Non-Prescription): ☐ Ca+ Channel Blocker ☐ Blood Pressure Medication ☐ Testosterone ☐ Proscar ☐ Marijuana ☐ Other Recreational Drugs ☑ N/A

Other Medication:

Medical Procedures: ☐ Vasectomy ☐ Vasectomy Repair ☐ Other Reproduction Tract Surgery ☐ Radiation Therapy ☐ Chemotherapy ☑ N/A

Recent Health: Have you been sick in the last three months? ☐ Yes ☑ No ☐ Fever?

Recent Health Comments:

Date Of Collection	12/13/2023	Collection Time		Received at lab by	Johnston, Laure	Time Sample Received	
Location of Collection	In Office	Container type	Sterile Cup/Sen	Days Abstinence	4 days		
Analyzed and Processed by	Sullivan, Leslie	Time of analysis		Method of Collection		Masturbation	
Complete Ejaculate?	Yes	Spillage Percent (%)		Analysis Tempature	☑ RT	Liquefaction (>2 cm)	**Complete by 60**
Viscosity	☐ < 2 cm ☐ > 2 cm	Agglutination– None	None	Characteristics	mod. debris		
Sample Color	☐ White ☐ Grey ☑ Yellow ☐ Clear ☐ Brown ☐ Red	Volume (<1.0mL)	**3.6 mL**				

Sperm Concentration	31.0 Mill/mL	Total Motility	52 %	Total sperm in ejaculate	111.60 Million
Total motile	58.03 Million	Progression	47	Non-Progression	6
Non-Motile	47				

Comments:

*WHO laboratory examination of human semen and sperm cervical mucus interaction, 5th edition, 2010.h

Time Complete: | Technician: Sullivan, Leslie | Secondary Witness: MC

Report Finalized by: Laura Mortensen on 12/13/2023 10:45 AM

End of **Final** report

Signature: _____