HEARING & VISION LOSS TEST REQUISITION

sema4
a Mount Sinai venture

SPECIMENS: 1428 Madison Ave., Rm AB2-25, New York, NY 10029
MAIL: One Gustave L. Levy Place, Box 1497, New York, NY 10029-6574
Phone: **800-298-6470** / Fax: 646-859-6870
Tax ID# 47-5349024/ CLIA# 33D2097541

ACCESSION NO.	DATE / /

PATIENT INFORMATION

LAST NAME	FIRST NAME
DATE OF BIRTH / /	BIOLOGICAL GENDER ☐ M ☐ F
PARTNER / SPOUSE LAST NAME	PARTNER / SPOUSE FIRST NAME
CLIENT MRN	PARTNER / SPOUSE DATE OF BIRTH / /
TELEPHONE	EMAIL
ADDRESS	CITY / STATE / ZIP

REFERRING PROVIDER INFORMATION

NAME	GENETIC COUNSELOR
ADDRESS	CLINIC / INSTITUTION
	TELEPHONE
	FAX

PROVIDER SIGNATURE OF CONSENT (REQUIRED): I certify that this patient (and/or their legal guardian, as necessary) has been informed of the benefits, risks, and limitations of the laboratory test(s) requested. I have answered this person's questions. I have obtained a signed informed consent from this patient or their legal guardian for this testing in accordance with applicable laws and regulations, including N.Y. Civil Rights Law Section 79-L, and will retain this consent in the patient's medical record.

SIGNATURE **DATE** / /

BILLING INFORMATION ■ Bill Clinic

POLICYHOLDER LAST NAME	POLICYHOLDER FIRST NAME	POLICYHOLDER DOB / /
INSURANCE CARRIER	INSURANCE ID	GROUP NO.
BILLING ADDRESS		
OTHER HEALTH COVERAGE (IDENTIFY)		

SELF-PAY: ☐ Credit Card ☐ Check

Make Checks Payable to:
Mount Sinai Genomics Inc., One Gustave L. Levy Place, Box 1497, New York, NY 10029

ASSIGNMENT AND RELEASE: I hereby authorize my insurance benefits be paid directly to the provider and I understand that I am financially responsible for uncovered services. I also authorize the release of any information required to process the claim. Billing inquiries, please call 800-298-6470, Option 3.

SIGNATURE DATE / /

CLINICAL INDICATIONS*

SPECIMEN TYPE
☐ Blood ☐ Saliva ☐ Amniotic Fluid
☐ CVS ☐ Other _____
☐ Cultured Cells Type_____

CLINICAL STATUS
☐ Affected ☐ Unknown (no screening/evaluation)
☐ Unaffected (all screening/evaluations(s) normal)

PURPOSE OF STUDY
☐ Diagnostic ☐ Carrier Testing
☐ Familial Follow-Up (Family Variant _____)
☐ Other: _____

Date/Time Specimen Drawn ____ /____ / ____ ____ : ____ AM PM

Date Specimen Sent ____ /____ / ____

PLEASE SUBMIT PEDIGREE IF AVAILABLE (SPACE ON BACK)

PATERNAL ANCESTRY:

MATERNAL ANCESTRY:

CONSANGUINITY? ☐ Yes ☐ No

PLEASE COMPLETE ALL CLINICAL QUESTIONS ON THE BACK PAGE

ICD10 Dx CODE(S)
☐ H90.5 (unspecified sensorineural hearing loss) ☐ H54.7 (unspecified visual loss)
☐ Other: _____

LABORATORY TEST(S) ORDERED

☐ **COMPREHENSIVE HEARING AND VISION LOSS PANEL (308 genes)** includes both Comprehensive Hearing Loss and Comprehensive Vision Loss Panels.
☐ **ADD-ON ULTRA-HIGH RESOLUTION HEARING AND VISION LOSS DEL/DUP ARRAY**

☐ **COMPREHENSIVE HEARING LOSS PANEL (92 genes)** includes *GJB2, GJB6, OTOA,* and *STRC* Del/Dup and Branchio-Oto-Renal Syndrome Panel, Usher Syndrome Panel, and Zellweger Syndrome Panel. The following 68 genes are in the Comprehensive Hearing Loss Panel but not in the subpanels:
☐*ACTG1,* ☐*AIFM1,* ☐*CACNA1D,* ☐*CCDC50,* ☐*CEACAM16,* ☐*CLDN14,* ☐*COCH,* ☐*COL11A2,* ☐*DFNA5,* ☐*DFNB59,* ☐*DIABLO,* ☐*DIAPH1,* ☐*EDN3,* ☐*EDNRB,* ☐*ESPN,* ☐*ESRRB,* ☐*EYA4,* ☐*GIPC3,* ☐*GPSM2,* ☐*GRHL2,* ☐*GRXCR1,* ☐*HGF,* ☐*ILDR1,* ☐*KARS,* ☐*KCNQ1,* ☐*KCNQ4,* ☐*LHFPL5,* ☐*LOXHD1,* ☐*LRTOMT,* ☐*MARVELD2,* ☐*MITF,* ☐*MSRB3,* ☐*MT-RNR1*(chrM:1494C>T & chrM:1555A>G only), ☐*MYH14,* ☐*MYH9,* ☐*MYO15A,* ☐*MYO3A,* ☐*MYO6,* ☐*OPA1,* ☐*OTOA* (CNV), ☐*OTOA* (NGS), ☐*OTOF,* ☐*OTOG,* ☐*OTOGL,* ☐*P2RX2,* ☐*PAX3,* ☐*PHYH,* ☐*POU3F4,* ☐*POU4F3,* ☐*PRPS1,* ☐*PTPRQ,* ☐*RDX,* ☐*SERPINB6,* ☐*SLC26A4,* ☐*SMPX,* ☐*SOX10,* ☐*STRC* (CNV), ☐*TBC1D24,* ☐*TECTA,* ☐*TIMM8A,* ☐*TMC1,* ☐*TMEM126A,* ☐*TMIE,* ☐*TMPRSS3,* ☐*TPRN,* ☐*TRIOBP,* ☐*TSPEAR,* ☐*WFS1*
☐**ADD-ON ULTRA-HIGH RESOLUTION HEARING LOSS DEL/DUP ARRAY**

☐**DFNB1-RELATED HEARING LOSS PANEL (2 genes)** includes ☐ *GJB2* (NGS) and ☐ *GJB2/GJB6* (CNV)

☐**BRANCHIO-OTO-RENAL SYNDROME PANEL (3 genes)** includes ☐*EYA1,* ☐*SIX1,* ☐*SIX5*
☐**ADD-ON ULTRA-HIGH RESOLUTION HEARING LOSS DEL/DUP ARRAY**

☐**USHER SYNDROME PANEL (11 genes)** includes ☐*ADGRV1,* ☐*CDH23,* ☐*CIB2,* ☐*CLRN1,* ☐*HARS,* ☐*MYO7A,* ☐*PCDH15,* ☐*USH1C,* ☐*USH1G,* ☐*USH2A,* ☐*WHRN*
☐**ADD-ON ULTRA-HIGH RESOLUTION HEARING LOSS DEL/DUP ARRAY**

☐**ZELLWEGER SYNDROME PANEL (9 genes)** includes ☐*PEX1,* ☐*PEX10,* ☐*PEX14,* ☐*PEX16,* ☐*PEX19,* ☐*PEX2,* ☐*PEX5,* ☐*PEX6,* ☐*PEX7*
☐**ADD-ON ULTRA-HIGH RESOLUTION HEARING LOSS DEL/DUP ARRAY**

☐ **COMPREHENSIVE VISION LOSS PANEL (250 genes)** includes Albinism, Hermansky-Pudlak Syndrome & Waardenburg Syndrome Panel, Developmental Eye Panel, Retinal Disease Panel, and Stickler & Cataract Panel. The following 33 genes are in the Comprehensive Vision Loss Panel but not in the subpanels: ☐*CABP4,* ☐*CDH3,* ☐*CLN3,* ☐*CLN5,* ☐*CLN6,* ☐*CLN8,* ☐*CTSD,* ☐*CYP1B1,* ☐*DNAJC5,* ☐*EFEMP1,* ☐*ELOVL4,* ☐*FRMD7,* ☐*FZD4,* ☐*GNAT1,* ☐*HESX1,* ☐*LYST,* ☐*MFSD8,* ☐*MTTP,* ☐*MYOC,* ☐*NYX,* ☐*PGK1,* ☐*PITX2,* ☐*PPT1,* ☐*RGS9,* ☐*RGS9BP,* ☐*RS1,* ☐*TGFBI,* ☐*TIMM8A,* ☐*TIMP3,* ☐*TPP1,* ☐*TREX1,* ☐*TSPAN12,* ☐*UBIAD1*
☐**ADD-ON ULTRA-HIGH RESOLUTION VISION LOSS DEL/DUP ARRAY**

☐**ALBINISM, HERMANSKY-PUDLAK SYNDROME, & WAARDENBURG SYNDROME PANEL (18 genes)** includes ☐*AP3B1,* ☐*BLOC1S6,* ☐*CACNA1F,* ☐*EDN3,* ☐*EDNRB,* ☐*GPR143,* ☐*HPS1,* ☐*HPS3,* ☐*HPS4,* ☐*HPS5,* ☐*HPS6,* ☐*MITF,* ☐*OCA2,* ☐*PAX3,* ☐*SLC45A2,* ☐*SOX10,* ☐*TYR,* ☐*TYRP1*
☐**ADD-ON ULTRA-HIGH RESOLUTION VISION LOSS DEL/DUP ARRAY**

☐**DEVELOPMENTAL EYE PANEL (21 genes)** includes ☐*BCOR,* ☐*BMP4,* ☐*FOXC1,* ☐*FOXE3,* ☐*FRAS1,* ☐*FREM1,* ☐*FREM2,* ☐*GRIP1,* ☐*HCCS,* ☐*KIF11,* ☐*MFRP,* ☐*NDP,* ☐*OTX2,* ☐*PAX6,* ☐*PITX3,* ☐*PLA2G5,* ☐*PXDN,* ☐*SMOC1,* ☐*SOX2,* ☐*STRA6,* ☐*VSX2*
☐**ADD-ON ULTRA-HIGH RESOLUTION VISION LOSS DEL/DUP ARRAY**

☐**RETINAL DISEASE PANEL (154 genes)** includes ☐*ABCA4,* ☐*ABHD12,* ☐*ADAM9,* ☐*ADGRV1,* ☐*AHI1,* ☐*AIPL1,* ☐*ALMS1,* ☐*ARL13B,* ☐*ARL6,* ☐*BBS1,* ☐*BBS10,* ☐*BBS12,* ☐*BBS2,* ☐*BBS4,* ☐*BBS5,* ☐*BBS7,* ☐*BBS9,* ☐*BEST1,* ☐*C2ORF71,* ☐*C5ORF42,* ☐*C8ORF37,* ☐*CA4,* ☐*CACNA1F,* ☐*CACNA2D4,* ☐*CAPN5,* ☐*CC2D2A,* ☐*CDH23,* ☐*CDHR1,* ☐*CEP164,* ☐*CEP290,* ☐*CEP41,* ☐*CERKL,* ☐*CHM,* ☐*CIB2,* ☐*CLRN1,* ☐*CNGA1,* ☐*CNGA3,* ☐*CNGB1,* ☐*CNGB3,* ☐*CNNM4,* ☐*CRB1,* ☐*CRX,* ☐*CYP4V2,* ☐*DHDDS,* ☐*EYS,* ☐*FAM161A,* ☐*FLVCR1,* ☐*FSCN2,* ☐*GNAT2,* ☐*GPR179,* ☐*GRK1,* ☐*GRM6,* ☐*GUCA1A,* ☐*GUCA1B,* ☐*GUCY2D,* ☐*HARS,* ☐*IFT140,* ☐*IMPDH1,* ☐*IMPG2,* ☐*INPP5E,* ☐*IQCB1,* ☐*KCNJ13,* ☐*KCNV2,* ☐*KIF7,* ☐*KLHL7,* ☐*LCA5,* ☐*LRAT,* ☐*LRIT3,* ☐*LZTFL1,* ☐*MAK,* ☐*MERTK,* ☐*MFRP,* ☐*MKKS,* ☐*MKS1,* ☐*MYO7A,* ☐*NMNAT1,* ☐*NPHP1,* ☐*NPHP3,* ☐*NPHP4,* ☐*NR2E3,* ☐*NRL,* ☐*OFD1,* ☐*OPA1,* ☐*OPN1SW,* ☐*PANK2,* ☐*PCDH15,* ☐*PDE6A,* ☐*PDE6B,* ☐*PDE6C,* ☐*PDE6G,* ☐*PDE6H,* ☐*PEX1,* ☐*PEX10,* ☐*PEX14,* ☐*PEX16,* ☐*PEX19,* ☐*PEX2,* ☐*PEX5,* ☐*PEX6,* ☐*PHYH,* ☐*PITPNM3,* ☐*PRCD,* ☐*PROM1,* ☐*PRPF3,* ☐*PRPF31,* ☐*PRPF6,* ☐*PRPF8,* ☐*PRPH2,* ☐*PRPS1,* ☐*RAB28,* ☐*RAX2,* ☐*RBP3,* ☐*RBP4,* ☐*RD3,* ☐*RDH12,* ☐*RDH5,* ☐*RGR,* ☐*RHO,* ☐*RIMS1,* ☐*RLBP1,* ☐*ROM1,* ☐*RP1,* ☐*RP1L1,* ☐*RP2,* ☐*RPE65,* ☐*RPGR,* ☐*RPGRIP1,* ☐*RPGRIP1L,* ☐*SAG,* ☐*SDCCAG8,* ☐*SEMA4A,* ☐*SLC24A1,* ☐*SNRNP200,* ☐*SPATA7,* ☐*TCTN1,* ☐*TCTN2,* ☐*TCTN3,* ☐*TMEM126A,* ☐*TMEM216,* ☐*TMEM231,* ☐*TMEM237,* ☐*TMEM67,* ☐*TOPORS,* ☐*TRIM32,* ☐*TRPM1,* ☐*TTC21B,* ☐*TTC8,* ☐*TULP1,* ☐*USH1C,* ☐*USH1G,* ☐*USH2A,* ☐*WDR19,* ☐*WHRN,* ☐*ZNF423*
☐**ADD-ON ULTRA-HIGH RESOLUTION VISION LOSS DEL/DUP ARRAY**

☐**STICKLER & CATARACT PANEL (41 genes)** includes ☐*ABHD12,* ☐*AGK,* ☐*BCOR,* ☐*BEST1,* ☐*CAPN5,* ☐*COL11A1,* ☐*COL11A2,* ☐*COL2A1,* ☐*COL9A1,* ☐*COL9A2,* ☐*CRX,* ☐*CRYAA,* ☐*CRYAB,* ☐*CRYBB1,* ☐*CRYBB3,* ☐*ERCC6,* ☐*EYA1,* ☐*FYCO1,* ☐*GCNT2,* ☐*GJA8,* ☐*GNPTG,* ☐*HSF4,*☐*JAG1,* ☐*KCNJ13,* ☐*LRP5,* ☐*MYH9,* ☐*NDP,* ☐*OAT,* ☐*OPA3,* ☐*OTX2,* ☐*PAX6,* ☐*PEX16,* ☐*PEX7,* ☐*PHYH,* ☐*PITX3,* ☐*PXDN,* ☐*RLBP1,* ☐*TDRD7,* ☐*VCAN,* ☐*VSX2,* ☐*WFS1*
☐**ADD-ON ULTRA-HIGH RESOLUTION VISION LOSS DEL/DUP ARRAY**

Patient Name: _____	Patient Name: _____	Patient Name: _____	Patient Name: _____

CLIENT SERVICES

HEARING LOSS CLINICAL INDICATIONS

AGE OF HEARING LOSS:

LATERALITY: ☐ Bilateral ☐ Unilateral

TYPE OF HEARING LOSS:
☐ Sensorineural ☐ Conductive ☐ Mixed
☐ Auditory neuropathy/dys-synchrony

PROGRESSION:
☐ Stable ☐ Progressive
☐ Fluctuating ☐ Unknown

SEVERITY (PTA): *Please attach audiogram if available.*

Left Ear: ☐ Mild (15-30dB) ☐ Moderate (31-50dB) ☐ Moderately-severe (51-70db) ☐ Severe (71-90db) ☐ Profound (>90dB)

Right Ear: ☐ Mild (15-30dB) ☐ Moderate (31-50dB) ☐ Moderately-severe (51-70db) ☐ Severe (71-90db) ☐ Profound (>90dB)

VISION LOSS CLINICAL INDICATIONS

AGE OF VISION LOSS:

LATERALITY: ☐ Bilateral ☐ Unilateral

TYPE OF VISION LOSS/SUSPECTED DIAGNOSIS:

PHENOTYPE (TO BE COMPLETED BY PHYSICIAN)

☐ Abnormal ERG	HP:0000512	☐ Intellectual disability	HP:0001249
☐ Absent ABR w/ cochlear microphonic	HP:0004463	☐ Keratoconus	HP:0000563
☐ Achromatopsia	HP:0011516	☐ Long QT	HP:0001657
☐ Aniridia	HP:0000526	☐ Macular degeneration	HP:0000608
☐ Balance problems	HP:0002141	☐ Mondini dysplasia	HP:0000376
☐ Branchial arch abnormality	HP:0009794	☐ Night Blindness	HP:0007642
☐ Cataracts	HP:0000518	☐ Nystagmus	HP:0000639
☐ Coloboma	HP:0000612	☐ Photophobia	HP:0000613
☐ Color blindness	HP:0007641	☐ Present OAEs	
☐ Cystoid macular edema	HP:0011505	☐ Renal abnormality	HP:0000077
☐ Delayed pupillary response	HP:0030211	☐ Retinal Detachment	HP:0000541
☐ Delayed walking	HP:0002194	☐ Retinitis pigmentosa	HP:0000510
☐ Dizziness/vertigo	HP:0002321	☐ Robin Sequence	HP:0000201
☐ Ear abnormalities	HP:0000377	☐ Skeletal abnormality	HP:0000924
☐ Ear tags	HP:0000384	☐ SNHL	HP:0000407
☐ EVA	HP:0011387	☐ Stapes fixation	HP:0000381
☐ Glaucoma	HP:0000501	☐ Tunnel vision	HP:0007994
☐ Heterochromia	HP:0001100	☐ Vivid blue eyes	HP:0000635
☐ Hirschprung	HP:0002251	☐ White forelock	HP:0002211

PEDIGREE AND ADDITIONAL NOTES

INFORMED CONSENT FOR GENETIC TESTING

I, _____, hereby request genetic testing for me/or my child (name of child if applicable) _____, which may include molecular, cytogenetic and/or biochemical analyses. I have received verbal and written information (please see https://sema4genomics.com/products/test-catalog/ for specific test-specific information sheet) from my physician or from a genetic counselor that described, in words that I understood, the nature of the genetic testing that I/my child am about to undergo.

I understand that a specimen(s), such as peripheral blood, saliva, cheek swab, dried blood spot, skin biopsy, amniotic fluid, chorionic villi and/or urine sample will be taken from me/my child. I understand that the samples will be used for determining if I/my child have a genetic disease, are carriers of a genetic disease or are more likely to develop a genetic disease or condition.

The nature of the genetic testing for (test name) _____ has been explained to me and the accuracy of the test and its risks and limitations have been detailed. I understand that although the likelihood of an incorrect diagnosis or a misinterpretation of the result is extremely small infrequent errors may occur. The likelihood of this occurring has been estimated to be less than 1%. I understand that a negative result reduces but does not eliminate the possibility that I/my child carry a mutation(s) in the gene(s) analyzed or in other gene(s) that are not included in the test.

No test will be performed on my sample other than the one(s) authorized by this consent and my doctor.

By signing this consent form, I agree that Sema4 may store, de-identify and use my/ my child's sample and information to support medical and academic research. Specimens from residents of New York will not be retained for more than 60 days after collection and will not be included in research studies unless I consent by initialing below. I understand that I may withdraw this consent at any time and that my/my child's specimen will be promptly destroyed.

_____ *Initial* **For residents of New York only,** I give consent to have my/my child's specimen anonymously used by Sema4 for scientific research related to genetic disease and stored for as long as the specimen is useful for such research purposes, not to exceed 10 years. I understand that I may withdraw this consent at any time and that my/my child's specimen will be promptly destroyed.

Sema4 may also give the de-identified information to its research partners and may submit this de-identified information to research databases for use in scientific and medical research, including scientific databases that are maintained by the federal government, such as a database kept by the National Institutes of Health ("NIH") (an agency of the federal government that funds research). I understand that I/my child will receive no compensation in connection with such research. If I prefer not to have any of my/my child's de-identified sample and health information used in research consistent with this consent, I may request this by contacting the laboratory.

I understand that this testing may yield results that are of unknown clinical significance and that parental or other relative's specimens may also be tested to determine whether a specific finding was inherited. In addition, incidental findings that are not related to the primary diagnosis may be identified in me/my child. An error in the diagnosis may occur if the true biological relationships of the family members involved are not as I have stated and this test may detect non-paternity.

I understand that the laboratory may wish to contact me/my child in the future for the following reasons: research purposes, the provision of general information about research findings, and/or the provision of information about the results of tests on my/my child's sample(s). I understand that I may notify the laboratory to opt out of such future contact.

The results of my/or my child's test will be explained to me by a genetic counselor or by my physician who will have the opportunity to discuss my results with a geneticist. I have had the opportunity to have all of my questions answered. If I am signing this form on behalf of a minor for whom I am the legal guardian, I am satisfied that I have received enough information to sign on his or her behalf.

I understand that this consent is being obtained in order to protect my right to have all of my questions answered before testing. I understand that the results of this testing will become part of my medical record and may only be disclosed to individuals who have legal access to this record or to individuals who I designate to receive this information.

_____ Signature of Person Being Tested (or guardian) / Date

_____ Signature of Witness / Date