GENETICS PRE-VISIT QUESTIONNAIRE

PATIENT NAME: **CURRENT AGE:**

BIOLOGICAL SEX: MALE FEMALE **DATE OF BIRTH:**

GENDER IDENTITY: MALE FEMALE NON-BINARY

(IF A MINOR) PATIENT'S MOTHER'S NAME :
PATIENT'S FATHER'S NAME:

REFERRED BY:

NAME:
ADDRESS:
TELEPHONE NUMBER:
FAX NUMBER:

PRIMARY CARE PHYSICIAN (IF DIFFERENT FROM REFERRING PHYSICIAN):

NAME:
ADDRESS:
TELEPHONE NUMBER:
FAX NUMBER:

WHY ARE YOU SEEKING A GENETIC EVALUATION?

WHEN DID THIS PROBLEM PRESENT?

WHICH OF THESE GENETIC TESTS HAVE BEEN PERFORMED ON YOU IN THE PAST?

- ❑ CHROMOSOME ANALYSIS (KARYOTYPE)
- ❑ CHROMOSOMAL MICROARRAY
- ❑ FISH TEST
- ❑ WHOLE EXOME SEQUENCING
- ❑ WHOLE GENOME SEQUENCING
- ❑ DNA METHYLATION ANALYSIS
- ❑ MUTATION ANALYSIS
- ❑ CARRIER SCREENING
- ❑ OTHER SPECIALIZED GENETIC STUDY
- ❑ *NONE OF THE ABOVE*

HAVE ANY OF THESE GENETIC TESTS BEEN PERFORMED ON A BLOOD RELATIVE? IF SO, PLEASE LIST THEIR FAMILIAL RELATIONSHIP TO YOU (E.G., MOTHER, FATHER, SISTER, COUSIN, ETC), NAME OF TEST, AND RESULT:

*****PLEASE FORWARD US RESULTS OF ALL PERTINENT GENETIC TESTS YOU OR YOUR RELATIVES HAVE HAD*****

WHICH OF THESE OTHER TESTS AND STUDIES HAVE YOU HAD?

- ❑ CT SCAN
- ❑ DEVELOPMENTAL ASSESSMENT
- ❑ ECHOCARDIOGRAM
- ❑ EEG (BRAIN)
- ❑ EKG (HEART)
- ❑ EYE/VISION EXAM
- ❑ HEARING EVALUATION
- ❑ MRI
- ❑ ORGAN BIOPSY
- ❑ OTHER BLOODWORK
- ❑ OTHER STUDIES (LIST HERE):
- ❑ SKIN BIOPSY
- ❑ ULTRASOUND
- ❑ X-RAYS

LIST RESULTS HERE (IF YOU NEED MORE SPACE PLEASE USE THE OTHER/COMMENTS SECTION AT THE END OF THIS FORM):

PRENATAL HISTORY

WHICH OF THE FOLLOWING APPLIED TO THE PATIENT'S MOTHER'S HEALTH DURING HER PREGNANCY WITH THE PATIENT? (CHECK OFF ALL THAT ARE APPLICABLE).

- ❑ ABNORMAL FETAL MOVEMENTS
- ❑ ALCOHOL USE
- ❑ BLEEDING
- ❑ CAFFEINE USE
- ❑ CHRONIC ILLNESS
- ❑ DRUG USE
- ❑ EXPOSURE TO CHEMICALS
- ❑ EXPOSURE TO X-RAYS
- ❑ FEVER
- ❑ FOLATE USE BEFORE CONCEPTION
- ❑ INFECTION
- ❑ MEDICATION USE
- ❑ OTHER PREGNANCY COMPLICATIONS
- ❑ PRENATAL VITAMIN USE
- ❑ TOBACCO USE
- ❑ *NONE OF THE ABOVE*

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PLEASE EXPLAIN IN MORE DETAIL THE ITEMS YOU HAVE CHECKED OFF:

WHAT PRENATAL TESTING WAS PERFORMED DURING THE PREGNANCY?

- ❑ AFP (OR TRIPLE SCREEN)
- ❑ AMNIOCENTESIS
- ❑ CHORIONIC VILLUS SAMPLING (CVS)
- ❑ GLUCOSE (SUGAR) TEST
- ❑ ULTRASOUND
- ❑ OTHER:

PLEASE DESCRIBE ANY ABNORMAL RESULTS:

BIRTH HISTORY

WAS THE PATIENT BORN:

- ❑ FULL-TERM
- ❑ PREMATURELY (NUMBER OF WEEKS:__________)

MODE OF DELIVERY:

- ❑ VAGINAL
- ❑ C-SECTION (WHY:_____________________________)

PRESENTATION:

- ❑ HEAD FIRST
- ❑ BOTTOM FIRST
- ❑ OTHER:

BIRTH WEIGHT: **BIRTH LENGTH:** **HEAD CIRCUMFERENCE:**

AT WHICH HOSPITAL (NAME, LOCATION, COUNTRY)?

APGAR SCORES, IF KNOWN:

BIRTH COMPLICATIONS, IF ANY:

LENGTH OF STAY IN NURSERY:

REASON FOR PROLONGED STAY, IF APPLICABLE:

MEDICAL HISTORY: *Has the patient had any of the following? If so, when?*

- ❑ ALLERGIES:_________________
- ❑ ANEMIA
- ❑ ANOREXIA
- ❑ ANXIETY DISORDER
- ❑ ASTHMA
- ❑ ATTENTION DEFICIT/ADHD
- ❑ BEDWETTING/SOILING
- ❑ BEHAVIOR PROBLEMS
- ❑ BIPOLAR DISORDER
- ❑ BLEEDING DISORDERS
- ❑ BLOOD TRANSFUSION (WHEN:____________________)
- ❑ CANCER
- ❑ CATARACTS
- ❑ CHLAMYDIA
- ❑ DEPRESSION
- ❑ DIABETES
- ❑ EMPHYSEMA
- ❑ EPILEPSY/SEIZURES
- ❑ FEEDING DIFFICULTIES
- ❑ GLAUCOMA
- ❑ GOITER
- ❑ GONORRHEA
- ❑ GOUT
- ❑ GROWTH PROBLEMS
- ❑ HEARING PROBLEMS
- ❑ HEART DISEASE
- ❑ HEPATITIS
- ❑ HERNIA (TYPE:_____________)
- ❑ HERPES (GENITAL)
- ❑ HIGH BLOOD PRESSURE
- ❑ HIGH CHOLESTEROL
- ❑ HIV POSITIVE
- ❑ HYPOTHYROIDISM
- ❑ HYPERTHYROIDISM
- ❑ INFECTIOUS DISEASE
- ❑ IMMUNOLOGICAL DISEASE
- ❑ KIDNEY DISEASE
- ❑ LEARNING DISABILITY
- ❑ LIVER DISEASE
- ❑ LOW BLOOD PRESSURE
- ❑ LOW CHOLESTEROL
- ❑ MANIC DISORDER
- ❑ MIGRAINES
- ❑ MISCARRIAGES
- ❑ MONONUCLEOSIS
- ❑ MOTOR DELAYS
- ❑ MULTIPLE SCLEROSIS
- ❑ MUSCULAR DYSTROPHY
- ❑ PACEMAKER
- ❑ PNEUMONIA
- ❑ PSYCHIATRIC DISORDER
- ❑ SCHIZOPHRENIA
- ❑ STD
- ❑ STROKE
- ❑ SUICIDE ATTEMPT
- ❑ SLEEPING PROBLEMS
- ❑ SPEECH DELAYS
- ❑ SYPHILIS
- ❑ THYROID DISEASE
- ❑ TUBERCULOSIS
- ❑ UNUSUAL HABITS
- ❑ ULCERS
- ❑ VISION PROBLEMS
- ❑ OTHER:
- ❑ NONE OF THE ABOVE

PLEASE EXPLAIN ANY ITEMS CHECKED OFF ABOVE:

DESCRIBE ANY CHRONIC MEDICAL ISSUES OR SIGNIFICANT ILLNESSES NOT LISTED ABOVE (IF NONE, WRITE "NONE"):

LIST PRIOR HOSPITALIZATIONS (HOSPITAL, DATE, DIAGNOSIS, TREATMENT) (IF NONE, WRITE "NONE"):

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LIST PRIOR SURGERIES (HOSPITAL, DATE, DIAGNOSIS) (IF NONE, WRITE "NONE"):

CURRENT PRESCRIBED OR OVER-THE-COUNTER MEDICATIONS THE PATIENT IS TAKING, INCLUDING DOSES:

LIST ALLERGIES TO MEDICATIONS AND TYPE OF REACTION:

DIETARY RESTRICTIONS IN PATIENT: **HOURS OF SLEEP PER NIGHT:**

IMMUNIZATIONS UP TO DATE? YES NO
HOW MANY COVID VACCINES HAVE YOU HAD (INCLUDE DATES)? 1ST DOSE: 2ND DOSE: 3RD DOSE:

PHYSICIANS INVOLVED IN THE PATIENT'S MEDICAL CARE (NAME/SPECIALITY/LAST VISIT/NEXT VISIT):

SOCIAL HISTORY:

- ❑ PATIENT LIVES WITH:
- ❑ ALCOHOL USE (HOW MUCH, HOW OFTEN):
- ❑ TOBACCO (HOW MANY CIGARETTES A DAY?)
- ❑ CAFFEINE INGESTION (HOW MUCH PER DAY?):
- ❑ OTHER SUBSTANCES

IF THE PATIENT IS A CHILD:

AT WHAT AGE WAS THE PATIENT FIRST ABLE TO DO THE FOLLOWING TASKS?

(IF YOU CANNOT REMEMBER WHEN, BUT YOUR CHILD CAN PERFORM THE TASK, JUST CHECK THE BOX. IF YOU KNOW THAT THE SKILL WAS REACHED LATER THAN NORMAL, WRITE "LATE"):

ASK FOR FOOD	SAY FIRST WORD
BABBLE	SAY MAMA/DADA
CRAWL	SIT UNSUPPORTED
CRAWL UP STAIRS	SMILE
ENJOY LOOKING AROUND	TELL HIS/HER NAME AND AGE
FINGERFEED	TELL STORIES
JUMP SKIP	TURN HEAD TO SOUND
LAUGH	USE A SPOON
LIFT CHEST WHILE ON STOMACH	USE A CUP
MANAGE STEPS WITHOUT HELP	USE TWO-WORD COMBINATIONS
NAME COLORS	WALK ALONE
PEDAL A TRICYCLE	WALK ALONG FURNITURE/ WALL
PLAY WITH OTHERS	WALK WITH HANDS HELD
POINT TO BODY PARTS	WAVE BYE-BYE
RAISE HEAD WHILE ON STOMACH	RECOGNIZE PARENTS
RECONGIZE STRANGERS	ROLL OVER
RUN	

LIST THE PATIENT'S GRADE LEVEL IN SCHOOL:

THE PATIENT IS ENROLLED IN:

- ❑ REGULAR CLASSES
- ❑ SPECIAL EDUCATION

THE PATIENT RECEIVES:

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- ❑ PHYSICAL THERAPY
- ❑ OCCUPATIONAL THERAPY
- ❑ SPEECH THERAPY
- ❑ INFANT STIMULATION
- ❑ ADAPTIVE PE
- ❑ OTHER THERAPY

HAS THE PATIENT BEEN REFERRED TO A REGIONAL CENTER:

❑ YES (WHICH ONE?_________________________________) ❑ NO

DO YOU HAVE ANY CONCERNS ABOUT THE PATIENT'S DEVELOPMENT OR SCHOOL PERFORMANCE?

IF THE PATIENT IS AN ADULT:

WHAT LEVEL OF EDUCATION HAS THE PATIENT ATTAINED?

DOES THE PATIENT HAVE DEVELOPMENTAL DELAY, LEARNING DISABILITY, OTHER COGNITIVE IMPAIRMENT, OR BEHAVIORAL DISORDER? IF SO, PLEASE DESCRIBE.

IS THE PATIENT EMPLOYED? YES NO

IF SO, WHAT IS THE OCCUPATION?

FAMILY HISTORY

ARE THERE ANY BLOOD RELATIVES WITH A SIMILAR PROBLEM AS THE PATIENT? YES NO

IF YES, WHO?

PATIENT'S MOTHER'S MEDICAL HISTORY

CURRENT AGE:
DATE OF BIRTH:
HEIGHT:
NUMBER OF TOTAL PREGNANCIES:
NUMBER OF LIVE BIRTHS:
NUMBER OF MISCARRIAGES:
NUMBER OF STILLBIRTHS:
MEDICAL PROBLEMS:
MEDICATIONS USED:

PATIENT'S FATHER'S MEDICAL HISTORY

CURRENT AGE:
DATE OF BIRTH:
HEIGHT:
MEDICAL PROBLEMS:
MEDICATIONS USED:

ARE THE PATIENT'S MOTHER AND FATHER BLOOD-RELATED (E.G., COUSINS?) YES NO

LIST THE PATIENT'S CHILDREN (IF ANY):

NAME	AGE	SEX	HEALTH

BROTHERS AND SISTERS OF PATIENT

(IF HALF-SIBLINGS, PLEASE NOTE MATERNAL OR PATERNAL) (ADD LINES IF NEEDED):

NAME	AGE	SEX	HEALTH	FULL OR HALF SIBLING

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DO THE PATIENT'S BROTHERS AND SISTERS HAVE KIDS?

ARE THEY HEALTHY?

GRANDPARENTS OF PATIENT

RELATIONSHIP	NAME	AGE	ETHNICITY *	HEALTH
MATERNAL GRANDMOTHER				
MATERNAL GRANDFATHER				
PATERNAL GRANDMOTHER				
PATERNAL GRANDFATHER				

*ETHNICITY: Mark if your grandparents or other ancestors are from any of these geographic regions or ethnic groups (relates to the epidemiology of some genetic diseases):

- ❑ African
- ❑ Ashkenazic Jewish
- ❑ Asian
- ❑ Caribbean
- ❑ Central American
- ❑ European
- ❑ Filipino
- ❑ French Canadian
- ❑ Hispanic
- ❑ Middle Eastern
- ❑ Middle Eastern Jewish
- ❑ Native American/Alaskan Native
- ❑ Pacific Islander
- ❑ Persian Jewish
- ❑ Sephardic Jewish
- ❑ South American
- ❑ Other:______________________

AUNTS AND UNCLES OF PATIENT

LIST THE <u>PATIENT'S MOTHER'S BROTHERS AND SISTERS</u> (IF HALF SIBLINGS, PLEASE NOTE MATERNAL OR PATERNAL) (ADD ADDITIONAL LINES IF NECESSARY)

NAME	AGE	SEX	HEALTH	FULL OR HALF SIBLING

LIST THE <u>PATIENT'S FATHER'S BROTHERS AND SISTERS</u> (IF HALF SIBLINGS, PLEASE NOTE MATERNAL OR PATERNAL) (ADD ADDITIONAL LINES IF NECESSARY)

NAME	AGE	SEX	HEALTH	FULL OR HALF SIBLING

IS THERE HISTORY IN THE PATIENT'S FAMILY OF:

- ❑ ATTENTION DEFICIT/HYPERACTIVITY
- ❑ AUTISM
- ❑ BIRTH DEFECTS
- ❑ CANCER
- ❑ DEVELOPMENTAL DELAY
- ❑ INFERTILITY
- ❑ MENTAL RETARDATION
- ❑ MISCARRIAGES
- ❑ NEWBORN/CHILDHOOD DEATHS
- ❑ PSYCHIATRIC DISORDERS
- ❑ STILLBIRTHS
- ❑ UNUSUAL FEATURES
- ❑ OTHER DISEASES:
- ❑ NONE OF THE ABOVE

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IF SO, PLEASE EXPLAIN IN MORE DETAIL:

REVIEW OF SYSTEMS

PATIENT HEIGHT:____**WEIGHT:**____

DOES THE PATIENT HAVE ANY OF THE FOLLOWING SYMPTOMS OR CONDITIONS? (IF SO, CHECK THEM OFF BELOW).

GENERAL
- ❑ FEVER
- ❑ CHILLS
- ❑ SWEATS
- ❑ CHANGE IN WEIGHT
- ❑ FATIGUE
- ❑ TEMPERATURE SENSITIVITY
- ❑ OTHER:
- ❑ NONE OF THE ABOVE

SKIN
- ❑ RASHES
- ❑ EASY BRUISING
- ❑ ECZEMA
- ❑ BIRTHMARKS
- ❑ LUMPS
- ❑ SKIN CHANGES
- ❑ NAIL CHANGES
- ❑ CHANGES IN MOLES
- ❑ OTHER:
- ❑ NONE OF THE ABOVE

EYES
- ❑ REDNESS
- ❑ DISCHARGE
- ❑ CHANGE IN VISION
- ❑ GLAUCOMA
- ❑ CATARACTS
- ❑ EYEGLASSES
- ❑ SENSITIVITY TO LIGHT
- ❑ OTHER:
- ❑ NONE OF THE ABOVE

EARS
- ❑ INFECTIONS
- ❑ RINGING
- ❑ HEARING IMPAIRMENT
- ❑ OTHER:
- ❑ NONE OF THE ABOVE

NOSE
- ❑ BLEEDING
- ❑ FREQUENT COLDS
- ❑ NASAL POLYPS
- ❑ OTHER:
- ❑ NONE OF THE ABOVE

MOUTH/THROAT
- ❑ PROBLEMS WITH TEETH
- ❑ PROBLEMS WITH GUMS
- ❑ FREQUENT SORE THROATS
- ❑ UNUSUAL VOICE
- ❑ OTHER:
- ❑ NONE OF THE ABOVE

NECK
- ❑ GOITER
- ❑ LUMPS
- ❑ PAIN
- ❑ THYROID PROBLEMS
- ❑ OTHER:
- ❑ NONE OF THE ABOVE

CHEST
- ❑ COUGH
- ❑ ASTHMA
- ❑ SNORING
- ❑ PAIN
- ❑ WHEEZING
- ❑ SHORTNESS OF BREATH
- ❑ SHORTNESS OF BREATH WHILE SLEEPING
- ❑ RESPIRATORY PROBLEMS
- ❑ PNEUMONIA
- ❑ OTHER:
- ❑ NONE OF THE ABOVE

HEART
- ❑ PALPITATIONS
- ❑ HEART MURMUR
- ❑ SHORTNESS OF BREATH WITH EXERTION
- ❑ OTHER:
- ❑ NONE OF THE ABOVE

DIGESTIVE
- ❑ BLOOD IN STOOLS
- ❑ CONSTIPATION
- ❑ DIARRHEA
- ❑ EXCESSIVE HUNGER
- ❑ FOOD INTOLERANCE
- ❑ LIVER DISEASE
- ❑ NAUSEA
- ❑ POOR APPETITE
- ❑ STOMACH ACHES
- ❑ SWALLOWING DIFFICULTY
- ❑ THIRST
- ❑ VOMITING
- ❑ OTHER:
- ❑ NONE OF THE ABOVE

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URINARY

- ☐ PAIN ON URINATION
- ☐ BLOOD IN URINE
- ☐ STRONG ODOR OF URINE
- ☐ FREQUENT URINATION
- ☐ BEDWETTING
- ☐ UNUSUAL URINE COLOR
- ☐ OTHER:
- ☐ NONE OF THE ABOVE

MUSCLE/BONE

- ☐ WEAKNESS
- ☐ LIMITATION OF MOVEMENT
- ☐ CRAMPS
- ☐ STIFFNESS
- ☐ JOINT PAIN
- ☐ OTHER:
- ☐ NONE OF THE ABOVE

NEUROLOGIC

- ☐ HEADACHE
- ☐ FAINTING
- ☐ TINGLING
- ☐ MOOD CHANGES
- ☐ SEIZURES
- ☐ SLEEP DISORDERS
- ☐ DIZZINESS
- ☐ TREMORS
- ☐ UNSTEADY GAIT
- ☐ TICS
- ☐ NUMBNESS
- ☐ SPEECH DISORDER
- ☐ OTHER:
- ☐ NONE OF THE ABOVE

PSYCHIATRIC

- ☐ LOSS OF INTEREST IN ACTIVITIES
- ☐ DIFFICULTY CONCENTRATING
- ☐ FEELING DEPRESSED/HOPELESS
- ☐ MOVING/SPEAKING MORE SLOWLY
- ☐ FEELING RESTLESS/FIDGETY
- ☐ WANTING TO HURT YOURSELF OR OTHERS
- ☐ OTHER:
- ☐ NONE OF THE ABOVE

GYN (FOR FEMALES)

- ☐ CURRENTLY PREGNANT? YES NO UNSURE
- ☐ IF YES, HOW MANY WEEKS?
- ☐ NUMBER OF PREGNANCIES:
- ☐ NUMBER OF LIVE BIRTHS:
- ☐ NUMBER OF MISCARRIAGES:
- ☐ NUMBER OF STILLBIRTHS:
- ☐ AGE AT FIRST PERIOD (MENARCHE):
- ☐ DATE OF LAST MENSTRUAL PERIOD:
- ☐ PROBLEMS WITH PERIODS (DESCRIBE):
- ☐ OTHER GYNECOLOGICAL PROBLEMS (DESCRIBE):
- ☐ PLAN ON BECOMING EGG DONOR
- ☐ OTHER:
- ☐ NONE OF THE ABOVE

UROLOGICAL (FOR MALES)

- ☐ LOW SPERM COUNT
- ☐ OTHER UROLOGICAL PROBLEMS
- ☐ MULTIPLE MISCARRIAGES IN FEMALE PARTNER
- ☐ PLAN ON BECOMING SPERM DONOR
- ☐ OTHER:
- ☐ NONE OF THE ABOVE

OTHER/ADDITIONAL COMMENTS

*THIS FORM WAS COMPLETED BY:*____ *DATE:*____________________

*REVIEWED BY PHYSICIAN:*____ *DATE:*____________________

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