Exome with CNV Evaluation Clinical History Form

Client account number: _____ Client name: _____

Primary patient name: _____

Primary patient DOB: _____ Patient phone number: _____

If ordering trio or duo testing, please provide the names and DOB of additional family members:

Name: _____ Relationship to Proband: _____ DOB: _____

Name: _____ Relationship to Proband: _____ DOB: _____

Proband information

Reason(s) for referral for exome sequencing, ie, what was the initial presenting symptom? _____

_____ Age of onset of initial presenting symptom: _____

Ethnicity (check all that apply):

☐ African or descended from the African continent ☐ Ashkenazi Jewish ☐ American Indian or Alaskan Native ☐ Asian ☐ Cajun/Creole
☐ Central or South American ☐ French Canadian ☐ Hispanic or Latino ☐ Middle Eastern ☐ Pacific Islander or Native Hawaiian
☐ Sephardic Jewish ☐ Western/Northern European ☐ Other (please specify): _____

Is there a family history of a similar or related disorder? ☐ Yes ☐ No

Consanguinity (related by blood, eg, parents related by blood)? ☐ Yes ☐ No ☐ Unsure If yes, please specify: _____

History of a bone marrow transplant: ☐ Yes* ☐ No
*If yes, please contact 1.866.GENE.INFO to speak to a Genomic Science Specialist before sending any samples.

Previous genetic testing

Chromosome analysis ☐ No results/not performed ☐ Yes/results: _____

Chromosomal microarray analysis ☐ No results/not performed ☐ Yes/results: _____

Other molecular studies, including prenatal testing: _____

Reporting of Secondary Findings (Physician Signature Required)

Secondary Findings Report: I have provided genetic counseling to the patient regarding the implications of receiving secondary findings. I have explained the potential benefits and limitations of receiving secondary findings, have answered the patient's questions, and have obtained the patient's consent regarding the reporting of secondary findings as indicated below. Check the appropriate option below to order reporting of medically actionable secondary findings recommended by ACMG. **If neither box is checked, secondary findings will not be reported.**

☐ **YES**, Please report pathogenic variants in genes determined to be medically actionable by the ACMG policy statement

☐ **NO**, Please DO NOT report pathogenic variants in genes determined to be medically actionable by the ACMG policy statement

SIGNATURE REQUIRED

Medical Professional's Signature: _____ Date _____

Patient Authorization to Use De-Identified Specimen (Patient Signature Required)

Test results are confidential and will only be reported as authorized by the patient or the patient's authorized representative or consistent with applicable state and federal law.

To promote medical understanding and develop better health insights, Quest Diagnostics requests the patient's permission to use the specimen *in a de-identified way* (without identifying the test subject) for research, educational studies, commercial purposes and/or publication. Your name or other personal identifying information will not be used in or linked to the results of any studies or publications.

You are not required to consent to any of these uses, and the decision to consent to the use of the specimen for *such purposes will not in any way affect processing or testing of the specimen, the test results or the services provided by Quest Diagnostics in connection with this testing*. Please indicate your choice regarding the use of the de-identified specimen by checking the line next to the appropriate option below.

☐ **YES**, I consent to the use of my de-identified specimen as described above.

☐ **NO**, I do not consent to the use my de-identified specimen as described above.

_____

Signature of Patient, Parent or Legally Authorized Representative Date

_____

Print Name Relationship

QuestDiagnostics.com

Clinical details

Date of last clinical exam: _____ Biological sex: ☐ Male ☐ Female ☐ Other (please specify): _____

Head circumference: _____ %tile Weight: _____ %tile Height: _____ %tile

Common diagnoses (please provide more information about these common diagnoses using the check boxes further below):

☐ Ambiguous genitalia ☐ Autism spectrum disorder ☐ Congenital heart defect ☐ Developmental delay ☐ Dysmorphic features

☐ Failure to thrive ☐ Hypotonia ☐ Metabolic acidosis ☐ Multiple congenital anomalies ☐ Seizures ☐ Structural brain abnormalities

Exome with CNV Evaluation Clinical History Form

Cancer

- ☐ Type of cancer: _____
- ☐ Age of diagnosis: _____
- ☐ Family history of cancer and affected relatives: _____

Cardiovascular

- ☐ Anemia
- ☐ Aortic root dilation
- ☐ Arrhythmia
- ☐ Atrial septal defect
- ☐ Bicuspid aortic valve
- ☐ Cardiomyopathy
- ☐ Coarctation of aorta
- ☐ EKG abnormality
- ☐ Mitral valve prolapse
- ☐ Patent ductus arteriosus
- ☐ Patent foramen ovale
- ☐ Teratology of Fallot
- ☐ Thrombocytopenia
- ☐ Thrombosis
- ☐ Tortuosity
- ☐ Truncus arteriosus
- ☐ Ventricular abnormality
- ☐ Ventricular septal defect

Craniofacial

- ☐ Bifid uvula
- ☐ Cleft lip
- ☐ Cleft palate
- ☐ Craniosynostosis
- ☐ Epicanthal folds
- ☐ Hypertelorism
- ☐ Hypotelorism
- ☐ Macrocephaly
- ☐ Microcephaly
- ☐ Micrognathia
- ☐ Nose abnormality
- ☐ Palpebral fissures
- ☐ Philtrum abnormality
- ☐ Teeth abnormality
- ☐ Tongue abnormality

Cognitive development

- ☐ ADD/ADHD
- ☐ Autism spectrum disorder
- ☐ Developmental delay
- ☐ Developmental regression
- ☐ Intellectual disability
  - ☐ Mild
  - ☐ Moderate
  - ☐ Severe
  - ☐ Profound
- ☐ Motor milestones delayed
- ☐ Speech delay

Ear & hearing

- ☐ Deafness
  - ☐ Acquired
  - ☐ Congenital
  - ☐ Bilateral
  - ☐ Unilateral
  - ☐ Conductive
  - ☐ Sensorineural
- ☐ Low-set ears
- ☐ Pinna abnormality
- ☐ Preauricular pit
- ☐ Preauricular skin tag

Endocrine

- ☐ Adrenal gland abnormality
- ☐ Adrenal insufficiency
- ☐ Cushing syndrome
- ☐ Diabetes insipidus
- ☐ Diabetes mellitus
- ☐ Growth hormone deficiency
- ☐ Hirsutism
- ☐ Immunologic abnormality
  Specify: _____
- ☐ Obesity
- ☐ Pancreatic insufficiency
- ☐ Parathyroid dysfunction
- ☐ Thyroid dysfunction

Eye defects & vision

- ☐ Amblyopia
- ☐ Aniridia
- ☐ Anophthalmia
- ☐ Blue sclerae
- ☐ Cataracts
  - ☐ Congenital
  - ☐ Postnatal
- ☐ Cherry red spot
- ☐ Coloboma
- ☐ Corneal abnormality
- ☐ Ectopia lentis
- ☐ Microphthalmia
- ☐ Nystagmus
- ☐ Ptosis
- ☐ Retinitis pigmentosa
- ☐ Strabismus
- ☐ Visual impairment
  - ☐ Blind
  - ☐ Cortical
  - ☐ Myopia

Gastrointestinal

- ☐ Anal malformation
- ☐ Constipation (chronic)
- ☐ Crohn's disease
- ☐ Diarrhea (chronic)
- ☐ Esophageal atresia
- ☐ Gastroesophageal reflux
- ☐ Gastroparesis
- ☐ Hepatic failure
- ☐ Hepatomegaly
- ☐ Hirschsprung disease
- ☐ Inflammatory bowel disease
- ☐ Intestinal pseudo-obstruction
- ☐ Pancreatitis
- ☐ Pyloric stenosis
- ☐ Splenomegaly
- ☐ Vomiting (episodic/cyclic)
- ☐ Tracheoesophageal fistula

Genitourinary

- ☐ Ambiguous genitalia
- ☐ Cryptorchidism
- ☐ Hypogonadism
- ☐ Hypospadias
- ☐ Kidney abnormality
  - ☐ Agenesis
  - ☐ Horseshoe
  - ☐ Partially duplicated
  - ☐ Polycystic
- ☐ Ovarian streak
- ☐ Polycystic ovarian syndrome
- ☐ Testicular abnormality
- ☐ Ureter abnormality
- ☐ Urethra abnormality

Hair & skin

- ☐ Albinism
- ☐ Blistering
- ☐ Cafe-au-lait spots

Hair & skin (continued)

- ☐ Hair
  - ☐ Alopecia
  - ☐ Brittle
  - ☐ Coarse
  - ☐ Hypopigmentation
- ☐ Hemangioma
- ☐ Hyperextensible skin
- ☐ Hyperpigmented macule
- ☐ Hypopigmented macule
- ☐ Hypertrichosis
- ☐ Ichthyosis
- ☐ Infections
- ☐ Lipoma
- ☐ Nail abnormality
- ☐ Neurofibroma
- ☐ Rash

Metabolic

- ☐ Acidosis
  - ☐ Lactic
  - ☐ Metabolic
- ☐ CSF lactate level (abnormal)
- ☐ Dicarboxylic aciduria
- ☐ Hyperammonemia
- ☐ Hyperglycemia
- ☐ Hypoglycemia
- ☐ Hyperphenylalaninemia
- ☐ Hypoammonemia
- ☐ Ketosis
- ☐ Organic aciduria
- ☐ Phosphokinase (abnormal)
- ☐ Plasma carnitine (abnormal)
- ☐ Serum creatine (abnormal)
- ☐ Serum pyruvate (abnormal)

Musculoskeletal

- ☐ Arthrogryposis
- ☐ Camptodactyly
- ☐ Contractures
- ☐ Fractures
- ☐ Hemihypertrophy
- ☐ Hyperlordosis
- ☐ Hypermobility
- ☐ Hypertonia
- ☐ Hypotonia
- ☐ Kyphosis
- ☐ Muscle atrophy
- ☐ Muscular dystrophy
- ☐ Myopathy
- ☐ Myotonia
- ☐ Oligodactyly
- ☐ Overgrowth
- ☐ Polydactyly
- ☐ Rib defects
- ☐ Scoliosis
- ☐ Short stature
- ☐ Skeletal dysplasia
- ☐ Spina bifida
- ☐ Syndactyly
- ☐ Talipes equinovarus
- ☐ Tall stature
- ☐ Vertebral anomalies

Neurologic

- ☐ Areflexia
- ☐ Ataxia
- ☐ Chorea
- ☐ Dystonia
- ☐ Epileptic encephalopathy
- ☐ Neuropathy
- ☐ Seizures
  - ☐ Absence
  - ☐ Atonic
  - ☐ Febrile
  - ☐ Generalized clonic
  - ☐ Generalized myoclonic
  - ☐ Generalized tonic
  - ☐ Generalized tonic-clonic
  - ☐ Infantile spasms
- ☐ Spasticity

Pre/perinatal history

- ☐ Conceived via artificial reproductive technology
- ☐ Congenital diaphragmatic hernia
- ☐ Cystic hygroma

Pre/perinatal history (continued)

- ☐ Encephalocele
- ☐ Increased nuchal translucency
- ☐ Intrauterine growth restriction
- ☐ Oligohydramnios
- ☐ Polyhydramnios
- ☐ Omphalocele
- ☐ Prematurity
- ☐ Teratogen exposure
  Specify: _____

Respiratory

- ☐ Apnea
- ☐ Asthma
- ☐ Bronchiectasis
- ☐ Hyperventilation
- ☐ Hypoventilation
- ☐ Pneumothorax
- ☐ Recurrent infections
- ☐ Respiratory failure
- ☐ Respiratory insufficiency

Structural brain abnormalities

- ☐ Aplasia/hypoplasia of the cerebellar vermis
- ☐ Aplasia/hypoplasia of the cerebellum
- ☐ Basal ganglia abnormality
- ☐ Brain atrophy
- ☐ Brainstem abnormality
- ☐ Cerebral dysmyelination
- ☐ Cerebral hypomyelination
- ☐ Cerebral white matter abnormality
- ☐ Corpus callosum abnormality
- ☐ Cortical dysplasia
- ☐ Encephalocele
- ☐ Holoprosencephaly
- ☐ Hydrocephalus
- ☐ Leukoencephalopathy
- ☐ Leukodystrophy
- ☐ Lissencephaly
- ☐ Neuronal migration abnormality
- ☐ Pachgyria
- ☐ Polymicrogyria
- ☐ Ventriculomegaly

Other

- ☐ Allergies (severe)
- ☐ Fever (episodic)
- ☐ Failure to thrive
- ☐ Heterotaxy
- ☐ Lethargy
- ☐ Organomegaly
- ☐ Pain (chronic)

Attach any imaging or laboratory results.

Questions? To speak with a Genomic Science Specialist, please call: **1.866.GENE.INFO**

Please fax or email the form to **1.949.668.7818** or **Preauthorization_neurology@QuestDiagnostics.com**