CONSTITUTIONAL GENETICS LABORATORY REQUISITION

DIVISION OF GENOME DIAGNOSTICS
at BC Children's & BC Women's Hospitals Facility Code L1050
4500 Oak Street Vancouver, BC V6H 3N1 www.genebc.ca
Molecular Genetics Tel: 604-875-2852, Fax: 604-875-2707
Cytogenetics Tel: 604-875-2304, Fax: 604-875-3601

***Yellow highlighted fields must be completed.**

SUNQUEST LABEL USE ONLY

DIVISION OF GENOME DIAGNOSTICS USE ONLY

Bill to → ☐ MSP ☐ PATIENT* ☐ OTHER*: _____ *Billing Form Required (www.genebc.ca)

ORDERING PRACTITIONER

ORDERING PRACTITIONER NAME | MSP #

ADDRESS

TELEPHONE | FAX

ALTERNATIVE CONTACT NAME | ALTERNATIVE CONTACT TELEPHONE

COPY TO PRACTITIONER

COPY TO PRACTITIONER NAME / ADDRESS | MSP #

COPY TO PRACTITIONER NAME / ADDRESS | MSP #

COPY TO PRACTITIONER NAME / ADDRESS | MSP #

PATIENT INFORMATION

PERSONAL HEALTH NUMBER | MEDICAL RECORD NUMBER | REFERRING CLINIC ID

LAST NAME OF PATIENT | FIRST NAME OF PATIENT

DOB YYYY MMM DD | SEX ☐ M ☐ F ☐ X | PATIENT ANCESTRY

PARTNER NAME | PARTNER PHN

PARTNER ANCESTRY | CONSANGUINITY ☐ Yes ☐ No ☐ UNK

☐ Test **impacts management** of an ongoing pregnancy

EDD YYYY MMM DD | PERSONAL HEALTH NUMBER (if other than above)

NAME & RELATIONSHIP (if other than above)

DIAGNOSIS/SUSPECTED DIAGNOSIS:

☐ Trisomy 21 ☐ Trisomy 18 ☐ Klinefelter ☐ Other:
☐ Trisomy 13 ☐ Turner

SAMPLE INFORMATION *for guidance: genebc.ca or eLabHandbook*

COLLECT EDTA, EXCEPT WHEN INDICATED BY # THEN COLLECT NAHEP

☐ EDTA Blood 2 mL ☐ NaHep Blood 3 mL
☐ DNA from: _____ Sample ID: _____
☐ Other: _____ Path #: _____

RELEVANT FAMILY HISTORY

Proband name/ PHN/Sample ID:
Proband relationship to this patient:
Proband diagnosis/result:

NON-SUNQUEST COLLECTION LAB LABEL USE ONLY

REASON FOR TESTING & CLINICAL INDICATIONS (pg 2) ARE REQUIRED

REASON FOR TESTING (CHECK ONE ONLY)

☐ **Confirmation of Diagnosis***
Has symptoms/signs

☐ **Presymptomatic Testing**
No symptoms/signs: risk of developing symptoms based on family history

☐ **Carrier Testing**
See genebc.ca for policy re:minors

* STAT newborn REQUIRES Suspected Dx (above)

TEST(S)

☐ Angelman Syndrome
☐ Ashkenazi Carrier Screening*
*Complete Supplemental Information Form
☐ Brugada Syndrome
☐ CADASIL
☐ Charcot-Marie-Tooth Type 1A (CMT1A)

Chromosome Analysis:
Comprehensive
☐ Breakage studies (Fanconi Anemia) #
☐ CMA (chromosome microarray)
☐ Karyotype #
Targeted
☐ Follow up variant assessment #
☐ Other #: _____

Cystic Fibrosis:
☐ Cystic Fibrosis
☐ CFTR-Related Disorder

Disorders of Sex Differentiation*:
*Restricted Ordering; see genebc.ca for details
☐ Androgen Insensitivity Syndrome
☐ Steroid-5-Alpha Reductase Deficiency
☐ Dystonia, early onset primary (DYT1)
☐ Dystrophinopathies (DMD, BMD)

FGFR3-Related Skeletal Dysplasia:
☐ Achondroplasia
☐ Hypochondroplasia
☐ Muenke Syndrome
☐ Thanatophoric Dysplasia

FMR1-Related Disorders:
☐ Fragile X Syndrome
☐ Fragile X-Associated Tremor Ataxia Syndrome (FXTAS)
☐ Fragile X-Associated Primary Ovarian Insufficiency *FSH required
☐ Friedreich Ataxia

Hemoglobin Disorders*:
*CBC & Hb HPLC/electrophoresis results required
☐ Alpha Thalassemia
☐ Beta Thalassemia
☐ Hemoglobin S, E, C
☐ Hereditary Neuropathy with Liability to Pressure Palsies (HNPP)
☐ Huntington Disease
☐ Hyperkalemic Periodic Paralysis
☐ Hypokalemic Periodic Paralysis
☐ Myotonic Dystrophy Type 1 (DM1)
☐ Myotonic Dystrophy Type 2 (DM2)
☐ Oculopharyngeal Muscular Dystrophy (OPMD)

Periodic Fever Syndromes:
☐ Familial Mediterranean Fever(FMF)
*Restricted Ordering; see genebc.ca for details
☐ Hyper IgD Syndrome (HIDS) *
☐ TNF receptor-associated periodic Syndrome (TRAPS) *
☐ Prader-Willi Syndrome
☐ Reference sample only (no report)
☐ Sensorineural Hearing Loss (GJB2/6)
☐ Spinal Muscular Atrophy (SMA)
☐ Spinal and Bulbar Muscular Atrophy (SBMA)
☐ Spinocerebellar Ataxia Panel (SCA 1, 2, 3, 6, 7)
☐ Transthyretin Amyloidosis (TTR)

Uniparental Disomy*:
*Parental reference samples required
☐ Chr 6 ☐ Chr 14
☐ Chr 7 ☐ Chr 15
☐ X-linked Ichthyosis (STS Deficiency)
☐ Zygosity *Medically indicated requests only
☐ Other: _____

PROVIDE CLINICAL INDICATIONS ON PAGE 2

DIVISION OF GENOME DIAGNOSTICS USE ONLY

DATE OF COLLECTION | TIME OF COLLECTION | COLLECTOR | **SIGNATURE OF PRACTITIONER** | **DATE SIGNED**

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PATIENT INFORMATION	
PERSONAL HEALTH NUMBER	
LAST NAME OF PATIENT	FIRST NAME OF PATIENT

CLINICAL FEATURES (CHECK ALL THAT APPLY)

Developmental/Psychiatric
- ☐ Autism Spectrum Disorder
- ☐ Developmental delay (specify domains):
  - ☐ Cognition
  - ☐ Fine motor ☐ Gross motor
  - ☐ Social
  - ☐ Speech/language
- ☐ Developmental regression
- ☐ Global developmental delay*
- ☐ Intellectual disability (specify):
  - ☐ Mild ☐ Moderate
  - ☐ Severe ☐ Profound
- ☐ Psychiatric disorder (specify in clinical notes)

*As defined by CPS (PMID: 30919832)

Growth Parameters
- ☐ Failure to thrive
- ☐ Hemihypertrophy
- ☐ Macrocephaly ☐ Microcephaly
- Weight for age: ☐ <3rd % ☐ >97th %
- Height for age: ☐ <3rd % ☐ >97th

Prenatal/Perinatal History
- ☐ Club foot
- ☐ Cystic hygroma
- ☐ Echogenic fetal bowel
- ☐ Gastroschisis
- ☐ Increased nuchal translucency
- ☐ Intrauterine growth restriction (IUGR)
- ☐ Meconium peritonitis
- ☐ Oligohydramnios
- ☐ Omphalocele
- ☐ Prematurity GA: _____
- ☐ Polyhydramnios

Auditory/Ophthalmologic
- ☐ Cataract
- ☐ Coloboma
- ☐ CPEO
- ☐ Hearing loss: ☐ Pre-lingual ☐ Post-lingual
- ☐ Optic neuropathy
- ☐ Ptosis
- ☐ Strabismus
- ☐ Visual impairment

Craniofacial/Skeletal
- ☐ Cleft lip/palate
- ☐ Craniosynostosis
- ☐ Facial dysmorphism (describe in clinical notes)
- ☐ Hypertelorism ☐ Hypotelorism
- ☐ Micrognathia or retrognathia
- ☐ Polydactyly
- ☐ Scoliosis
- ☐ Syndactyly

Cardiovascular
- ☐ Aortic dilatation/dissection
- ☐ Arrhythmia/Conduction defect:
  - ☐ Brugada type 1 ECG pattern
  - ☐ Prolonged QTc interval
- ☐ Cardiac amyloidosis
- ☐ Cardiomyopathy:
  - ☐ Dilated
  - ☐ Hypertrophic
  - ☐ Non-compaction
- ☐ Congenital heart defect:
  - ☐ Atrial septal defect (ASD)
  - ☐ Bicuspid aortic valve
  - ☐ Coarctation of aorta
  - ☐ Patent ductus arteriosus (PDA)
  - ☐ Tetralogy of Fallot
  - ☐ Ventricular Septal Defect (VSD)
- ☐ Syncopy

Cutaneous
- ☐ Café-au-lait macules
- ☐ Connective tissue abnormality
- ☐ Hyperpigmentation of the skin
- ☐ Hypopigmentation of the skin
- ☐ Ichthyosis

Endocrine
- ☐ Diabetes mellitus:
  - ☐ Type 1
  - ☐ Type 2
- ☐ Elevated FSH: _____
- ☐ Hypogonadism
- ☐ Precocious puberty
- ☐ Primary amenorrhea
- ☐ Transient neonatal diabetes mellitus

Gastrointestinal
- ☐ Inguinal hernia
- ☐ Meconium ileus
- ☐ Pancreatitis
- ☐ Pancreatic insufficiency
- ☐ Tracheoesophageal fistula

Genitourinary
- ☐ Ambiguous genitalia
- ☐ Congenital Absence of the Vas Deferens (CAVD)
- ☐ Cryptorchidism
- ☐ Hypospadias
- ☐ Hydronephrosis
- ☐ Obstructive azoospermia
- ☐ Primary ovarian insufficiency; FSH: _____
- ☐ Renal anomalies (describe in clinical notes)

Neurological
- ☐ Dementia
- ☐ Dysarthria
- ☐ Dysphagia
- ☐ Encephalopathy
- ☐ Headaches
- ☐ Hemiplegia
- ☐ Infantile spasms
- ☐ Migraines
- ☐ Seizure (specify type in clinical notes, if known)
- ☐ Stroke ☐ Stroke-like episodes

Neuromuscular
- ☐ Arthrogryposis
- ☐ Ataxia
- ☐ Chorea
- ☐ Calf muscle pseudohypertrophy
- ☐ Carpal tunnel syndrome
- ☐ Contracture
- ☐ Dystonia
- ☐ Fasciculations
- ☐ Hypertonia
- ☐ Hypotonia
- ☐ Muscular atrophy:
  - ☐ Proximal ☐ Distal
- ☐ Muscular dystrophy
- ☐ Muscle weakness:
  - ☐ Proximal ☐ Distal
- ☐ Myoclonus
- ☐ Myopathy
- ☐ Myotonia
- ☐ Neuropathy:
  - ☐ Motor ☐ Sensory
  - ☐ Focal ☐ Poly
- ☐ Pes cavus
- ☐ Spasticity
- ☐ Torticollis
- ☐ Tremor

Structural brain abnormalities
- ☐ Abnormal myelination
- ☐ Agenesis of the corpus callosum
- ☐ Cerebellar atrophy
- ☐ Cortical dysplasia
- ☐ Hemimegalencephaly
- ☐ Holoprosencephaly
- ☐ Hydrocephalus
- ☐ Leukodystrophy
- ☐ Lissencephaly
- ☐ Pachygyria
- ☐ Polymicrogyria
- ☐ Ventriculomegaly
- ☐ White matter hyperintensities

ADDITIONAL RELEVANT INFORMATION (CLINICAL NOTES, FAMILY HISTORY, PEDIGREE)

Print Form Reset Form