CONSTITUTIONAL GENETICS LABORATORY REQUISITION CWGG_REQ_0000 2026/11/17

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

Patient Education:
BC.mygeneticsadviser.com

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

ORDERING PRACTITIONER: ADDRESS, PHONE, MSP PRACTITIONER NUMBER
PRACTITIONERS with multiple clinic sites indicate address results to be sent

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

PERSONAL HEALTH NUMBER	MEDICAL RECORD NUMBER	REFERRING CLINIC ID	If LOCUM above indicate for which PRACTITIONER and include MSP #

LAST NAME OF PATIENT	FIRST NAME OF PATIENT	Copy to PRACTITIONER/MSP Practitioner Number & Clinic Name/Address

DOB YYYY MMM DD	SEX	PATIENT ANCESTRY	Copy to PRACTITIONER/MSP Practitioner Number & Clinic Name/Address
	☐ M ☐ F ☐ X ☐ UNK		

PARTNER NAME	PARTNER PHN	Copy to PRACTITIONER/MSP Practitioner Number & Clinic Name/Address

PARTNER ANCESTRY	CONSANGUINITY	Copy to PRACTITIONER/MSP Practitioner Number & Clinic Name/Address
	☐ YES ☐ NO ☐ UNK	

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

NON-PRACTITIONER CONTACT (FIRST NAME ONLY), PHONE

EDD YYYY MMM DD	NAME OF PREGNANT INDIVIDUAL & RELATIONSHIP (if other than above)	PERSONAL HEALTH NUMBER OF PREGNANT INDIVIDUAL (if other than above)

DIAGNOSIS:

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

SAMPLE INFORMATION *for guidance: genebc.ca or eLab Handbook*

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:

COLLECTION LAB LABEL

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

REASON FOR TESTING (CHECK ONE ONLY)

☐ **Confirmation of Diagnosis***
*COMPLETE Diagnosis + Clinical features
Has symptoms / signs

☐ **Presymptomatic Testing***
*COMPLETE relevant family history
Family Hx, Predictive, No symptoms / signs

☐ **Carrier Testing***
*COMPLETE relevant family history

TEST(S)

☐ Angelman Syndrome
☐ Ashkenazi Jewish Carrier Screening*
*COMPLETE Supplemental Information Form
☐ Brugada Syndrome
☐ CADASIL
☐ Charcot-Marie-Tooth Type 1A
Chromosome Analysis:
Comprehensive
☐ Breakage studies (Fanconi Anemia)
☐ CMA (chromosome microarray)
☐ Karyotype #
Targeted
☐ Follow up variant assessment #
☐ Other #: _____
Cystic Fibrosis:
☐ Cystic Fibrosis
☐ CFTR-Related Disorder
☐ Disorder of Sexual Development*:
*Restricted ordering
Androgen Insensitivity Syndrome
Steroid-5-Alpha Reductase Deficiency
☐ Dystonia, early onset primary (DYT1)
☐ Dystrophinopathies (DMD, BMD)
☐ FGFR3-Related Disorder:
Achondroplasia
Hypochondroplasia
Muenke Syndrome
Thanatophoric Dysplasia
FMR1-Related Disorder:
☐ Fragile X Syndrome
☐ Fragile X-Associated Tremor Syndrome Ataxia Syndrome (FXTAS)
☐ Fragile X-Associated Primary Ovarian Insufficiency *FSH REQUIRED
☐ Friedreich Ataxia
Genome Wide Sequencing *Restricted ordering
☐ Trio ☐ Singleton
☐ Duo ☐ + mtDNA
Hemoglobin Disorder*:
*CBC & Hb HPLC/electrophoresis REQUIRED
☐ Alpha Thalassemia
☐ β-Globin-Related Hemoglobinopathy
☐ Hereditary Neuropathy with Liability to Pressure Palsies (HNPP)
☐ Huntington Disease
☐ Hyper-/Hypo- kalemic Periodic Paralysis
☐ Myotonic Dystrophy Type 1 (DM1)
☐ Myotonic Dystrophy Type 2 (DM2)
☐ Oculopharyngeal Muscular Dystrophy (OPMD)
☐ Other:
_____
☐ Periodic Fever Syndrome:
Cryopyrin-Associated Periodic Syndrome (CAPS)
Familial Mediterranean Fever (FMF)
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 only

PROVIDE CLINICAL INDICATIONS ON PAGE 2

GENOME DIAGNOSTICS USE ONLY

DIVISION OF GENOME DIAGNOSTICS USE ONLY

DATE OF COLLECTION	TIME OF COLLECTON	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
- ☐ Atypical behaviour; specify in clinical notes
- ☐ Autism
- ☐ Developmental regression
- ☐ Global developmental delay*CPS definition (PMID: 30919832)
- ☐ Intellectual disability; specify:
  - ☐ Mild ☐ Moderate
  - ☐ Severe ☐ Profound
- ☐ Neurodevelopmental delay (isolated or <2SD); specify:
  - ☐ Cognitive skills
  - ☐ Fine motor ☐ Gross motor
  - ☐ Social
  - ☐ Speech/language

Growth Parameters
- ☐ Failure to thrive
- ☐ Hemihypertrophy
- ☐ Macrocephaly ☐ Microcephaly
- ☐ Short stature (<2SD) ☐ Tall stature (>2SD)
- ☐ Increased body weight ☐ Decreased body weight

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

Auditory/Ophthalmologic
- ☐ Cataract
- ☐ Coloboma
- ☐ External ophthalmoplegia
- ☐ Hearing impairment, specify:
  - ☐ Pre-lingual ☐ Post-lingual
- ☐ Optic neuropathy
- ☐ Ptosis
- ☐ Strabismus
- ☐ Visual impairment

Craniofacial/Skeletal
- ☐ Orofacial cleft
- ☐ Craniosynostosis
- ☐ Dysmorphic facies; describe in clinical notes
- ☐ Hypertelorism ☐ Hypotelorism
- ☐ Micrognathia ☐ Retrognathia
- ☐ Polydactyly ☐ Syndactyly
- ☐ Scoliosis

Cardiovascular
- ☐ Abnormal heart morphology, specify:
  - ☐ Atrial septal defect (ASD)
  - ☐ Bicuspid aortic valve
  - ☐ Coarctation of aorta
  - ☐ Patent ductus arteriosus (PDA)
  - ☐ Tetralogy of Fallot
  - ☐ Ventricular Septal Defect (VSD)
- ☐ Aortic dilatation / aneurysm
- ☐ Aortic dissection
- ☐ Arrhythmia:
  - ☐ with Abnormal EKG
- ☐ ATTR cardiac amyloidosis
- ☐ Cardiac conduction abnormality:
  - ☐ with abnormal EKG
  - ☐ with prolonged QTc interval
- ☐ Non-compaction cardiomyopathy
- ☐ Syncope

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

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

Gastrointestinal
- ☐ Inguinal hernia
- ☐ Meconium ileus
- ☐ Pancreatitis
- ☐ Exocrine pancreatic insufficiency
- ☐ Tracheoesophageal fistula

Genitourinary
- ☐ Ambiguous genitalia
- ☐ Absent vas deferens
- ☐ Cryptorchidism
- ☐ Hypospadias
- ☐ Hydronephrosis
- ☐ Kidney abnormality (describe in clinical note)
- ☐ Obstructive azoospermia
- ☐ Primary ovarian insufficiency; FSH: _____

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

Neuromuscular
- ☐ Arthrogryposis multiplex congenita
- ☐ Ataxia
- ☐ Chorea
- ☐ Calf muscle pseudohypertrophy
- ☐ Carpal tunnel syndrome
- ☐ Dystonia
- ☐ Fasciculations
- ☐ Hypertonia
- ☐ Hypotonia
- ☐ Joint contracture
- ☐ Skeletal muscular atrophy; specify:
  - ☐ Distal amyotrophy ☐ Proximal amyotrophy
- ☐ Muscular dystrophy
- ☐ Muscle weakness; specify:
  - ☐ Distal ☐ Proximal
- ☐ Myoclonus
- ☐ Myopathy
- ☐ Myotonia
- ☐ Peripheral neuropathy; specify:
  - ☐ Motor ☐ Sensory ☐ Autonomic
- ☐ 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, HPO Terms)

HPO#	Associated term	HPO#	Associated term
e.g. HP:0001631	e.g. Atrial septal defect		

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