Whole Exome Requisition Form

This requisition form, and consent forms in other languages, can be printed from **www.blueprintgenetics.com**, where test codes and detailed descriptions on tests and analysis types are also available.

Blueprint Genetics

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Promotion/Contract Code: .....

REQUIRED FIELDS ARE MARKED WITH AN ASTERISK (*)

TEST SELECTION*

☐ **Whole Exome Plus** ☐ **Whole Exome Family Plus^**	^Sample and Informed Consent is needed from all family members included in the Family test.
☐ **Expand to Exome^** **Specify the previous order ID:**	^ Expand your patient's previous test done at Blueprint Genetics (panel, single gene) to Whole Exome Plus or Whole Exome Family Plus. You will be contacted if new sample is needed.
Sample Type: ☐ **Blood** ☐ **Saliva** ☐ **DNA, source:**.....	**Sample Collection Date:**

ORDERING HEALTH CARE PROFESSIONAL INFORMATION

Name and Full Address:*	**Institution:***
	Email:*
	NPI# (US only):
	Phone:
	Fax:
☐ **Mail Results** ☐ **Fax Results** ☑ **Nucleus**	Results will always be available on our online reporting system at nucleus.blueprintgenetics.com

Filtered variant results files and raw data files can be provided on separate request. Please contact support@blueprintgenetics.com.

SHARE RESULTS WITH

Name:		**Role/Title:**	
Email:		**Street Address:**	
City:	**State:**	**Zip/Post Code:**	**Country:**
Phone:	**Fax:**	☐ **Fax Results** ☐ **Mail Results** ☐ **Nucleus**	Results can be shared within the same hospital on our ordering portal, Nucleus.

PATIENT INFORMATION

First Name:*	**Last Name:***	**DOB:***	**MRN/SSN:**
Street Address:			
City:	**State:**	**Zip/Post Code:**	**Country:**
Phone:	**Email:**		

FAMILY MEMBER 1 INFORMATION (FILL ONLY IF WHOLE EXOME FAMILY PRODUCT IS ORDERED)

First Name:*	**Last Name:***	**DOB:***	**MRN/SSN:**
Relationship to Patient:*		**Phenotype Description:**	
Is Family Member 1 Affected With the Same Phenotype as Patient:* ☐ Yes ☐ No ☐ Partially ☐ Uncertain			

FAMILY MEMBER 2 INFORMATION (FILL ONLY IF WHOLE EXOME FAMILY PRODUCT IS ORDERED)

First Name:*	**Last Name:***	**DOB:***	**MRN/SSN:**
Relationship to Patient:*		**Phenotype Description:**	
Is Family Member 2 Affected With the Same Phenotype as Patient:* ☐ Yes ☐ No ☐ Partially ☐ Uncertain			

If you wish to send more than two family members for Whole Exome Family test, please contact our support@blueprintgenetics.com.

PATIENT HISTORY (DETAILED CLINICAL INFORMATION IS ESSENTIAL FOR ACCURATE INTERPRETATION OF RESULTS)

Sex:* ☐ Male ☐ Female ☐ Unknown/uncertain	**Ethnicity:**
ICD-10 Codes:* (required for patient insurance – US only)	
Has the Patient Died? ☐ Yes ☐ No	**Hospital Status:** ☐ Inpatient ☐ Outpatient ☐ Non-hospital patient
Has the Patient or Either of the Family Members Had a Bone Marrow Transplant?* ☐ No ☐ Yes, specify who:.....	**Has the Patient or Either of the Family Members Had Blood Transfusions in the Past Two Weeks?*** ☐ No ☐ Yes, specify who:.....
Describe All Clinical Findings * (Attach possible supportive material.) Variants are reported based on the clinical information provided, therefore detailed phenotypic and clinical information increases the likelihood of a diagnosis.	
Family History (e.g. parental consanguinity, attach pedigree if available):	
Previous Testing With Normal Results:	**Previous Testing With Abnormal Results:**
Please specify genes of interest:	**Please specify suspected differential diagnosis (if applicable):**

CLINICAL FEATURES CHECKLIST

Perinatal History
- ☐ Cystic hygroma
- ☐ Increased nuchal translucency
- ☐ Intrauterine growth restriction
- ☐ Oligohydramnios
- ☐ Polyhydramnios
- ☐ Prematurity
- ☐ Other:.....

Cardiovascular
- ☐ Angioedema
- ☐ Aortic dilatation
- ☐ Arrhythmia / conduction defect
- ☐ Atrial septal defect
- ☐ Cardiomyopathy
- ☐ Coarctation of aorta
- ☐ Hypoplastic left heart
- ☐ Malformation of heart and/or great vessels
- ☐ Stroke
- ☐ Tetralogy of Fallot
- ☐ Ventricular septal defect
- ☐ Other:.....

Dermatological
- ☐ Blistering
- ☐ Connective tissue abnormality
- ☐ Hair abnormality
- ☐ Pigmentation abnormality
- ☐ Ichthyosis
- ☐ Skin tumors
- ☐ Nail abnormality
- ☐ Other:.....

Endocrinological
- ☐ Diabetes mellitus
- ☐ Hyperparathyroidism
- ☐ Hypoparathyroidism
- ☐ Hyperthyroidism
- ☐ Hypothyroidism
- ☐ Paraganglioma
- ☐ Pheochromocytoma
- ☐ Other:.....

Gastroenterological
- ☐ Constipation
- ☐ Chronic diarrhea
- ☐ Chronic intestinal pseudo-obstruction
- ☐ Elevated transaminases
- ☐ Gastroesophageal reflux
- ☐ Gastroschisis
- ☐ Hepatic failure
- ☐ Hirschsprung disease
- ☐ Pyloric stenosis
- ☐ Recurrent vomiting
- ☐ Tracheoesophageal fistula
- ☐ Other:.....

Hematological and Immunological
- ☐ Anemia
- ☐ Coagulation disorder
- ☐ Immunodeficiency
- ☐ Myelofibrosis
- ☐ Neutropenia
- ☐ Pancytopenia
- ☐ Thrombocytopenia
- ☐ Other:.....

Malformations – Brain
- ☐ Abnormalities of basal ganglia
- ☐ Agenesis of the corpus callosum
- ☐ Brain atrophy
- ☐ Cortical dysplasia
- ☐ Hemimegalencephaly
- ☐ Heterotropia
- ☐ Holoprosencephaly
- ☐ Hydrocephalus
- ☐ Lissencephaly
- ☐ Macrocephaly
- ☐ Microcephaly
- ☐ Periventricular leukomalacia
- ☐ Other:.....

Malformations - Skeletal and Other
- ☐ Cleft lip / palate
- ☐ Club foot / feet
- ☐ Contractures
- ☐ Craniosynostosis
- ☐ Dysmorphic features
- ☐ Ear malformation
- ☐ Fractures
- ☐ Limb anomaly
- ☐ Overgrowth
- ☐ Polydactyly
- ☐ Scoliosis
- ☐ Short stature
- ☐ Syndactyly
- ☐ Vertebral anomaly
- ☐ Other:.....

CLINICAL FEATURES CHECKLIST

Metabolic
- ☐ Abnormal creatine phosphokinase
- ☐ Elevated alanine
- ☐ Elevated puryvate
- ☐ Failure to thrive
- ☐ Ketosis
- ☐ Lactic acidosis
- ☐ Organic aciduria
- ☐ Other:.....

Nephrological
- ☐ Hydronephrosis
- ☐ Kidney malformation
- ☐ Renal agenesis or dysgenesis
- ☐ Renal tubulopathy
- ☐ Other:.....

Neurodevelopmental
- ☐ ADHD
- ☐ Autism spectrum disorder
- ☐ Developmental delay
- ☐ Developmental regression
- ☐ Encephalopathy
- ☐ Fine motor delay
- ☐ Gross motor delay
- ☐ Hearing loss
- ☐ Intellectual disability
- ☐ Learning disability
- ☐ Obsessive-compulsive disorder
- ☐ Psychiatric symptoms
- ☐ Recurrent headache
- ☐ Seizures
- ☐ Speech delay
- ☐ Other:.....

Neuromuscular
- ☐ Ataxia
- ☐ Chorea
- ☐ Dystonia
- ☐ Hypotonia
- ☐ Hypertonia
- ☐ Muscle weakness
- ☐ Muscular dystrophy
- ☐ Neuropathy
- ☐ Spasticity
- ☐ Other:.....

Ophthalmological
- ☐ Abnormal eye movement
- ☐ Abnormal vision
- ☐ Blindness
- ☐ Cataracts
- ☐ Coloboma
- ☐ CPEO
- ☐ Optic atrophy
- ☐ Ptosis
- ☐ Retinitis pigmentosa
- ☐ Other:.....

Reproductive System Abnormalities
- ☐ Ambiguous genitalia
- ☐ Cryptochidism
- ☐ Hypogonadism
- ☐ Hypospadias
- ☐ Infertility
- ☐ Undescended testis
- ☐ Other:.....

Tumors / Malignancies
- ☐ Adenomatous polyposis
- ☐ Brain tumor
- ☐ Breast cancer
- ☐ Colorectal cancer
- ☐ Leukemia
- ☐ Lung cancer
- ☐ Melanoma
- ☐ Other:.....

BILLING INFORMATION*

☐ **INSURANCE BILLING**

Include copy of both sides of the insurance card. The insurance provider usually requests a letter of medical necessity after submission of the claim.

Insurance Company:

Insurance ID #:

Group #:

Patient Relation to Policy Holder:
☐ Self ☐ Spouse ☐ Child ☐ Other

Policy Holder Name:

Policy Holder DOB:

☐ **INSTITUTIONAL BILLING**

Facility Name:

Street Address:

City: **State:**

Zip/Post Code: **Country:**

Contact Person:

Phone: **Fax:**

☐ **PATIENT PAYMENT**

Patient payment is by credit card. The payment process begins with the patient receiving a link with payment details to the email address filled out below. The sample goes to analysis once the payment has been collected by Blueprint Genetics.

Please contact billing.us@blueprintgenetics.com or (650) 452-9340 ext 1 if you wish to discuss alternative payment options.

Email:

Name:

Phone:

ORDERING HEALTH CARE PROFESSIONAL SIGNATURE

I have discussed the Informed Consent for Whole Exome Sequencing with the patient or their legal guardian and possible family members included in the test (Whole Exome Family products). I have obtained any other consent from the patient and family members that is required under the laws of my country/state and/or federal laws. I certify that the test ordered is medically necessary for the diagnosis or detection of a disease, illness, impairment, symptom, syndrome, or disorder. The results of this test will be used in the medical management of the patient and/or genetic counseling of the patient and family member(s). I have read and understood the General Terms of Service.

Signature:* **Date:***

HEALTH CARE PROFESSIONAL SIGNATURE REQUIRED FOR PROCESSING

GENERAL TERMS

By placing the order the Customer accepts Blueprint Genetics' General Terms. Blueprint Genetics reserves the right to amend its General Terms, of which the latest version shall always be applied. The latest version can be found at https://blueprintgenetics.com/general-terms/

The Informed Consent for Whole Exome Sequencing is available in different languages (Danish, English, Finnish, French, German, Hungarian, Italian, Norwegian, Polish, Spanish and Swedish) at www.blueprintgenetics.com/how-to-order/. **When ordering a Whole Exome Family product, please print out a separate Informed Consent for each family member.**

INFORMED CONSENT

Whole Exome Sequencing

About Whole Exome sequencing

Reaching a correct genetic diagnosis in a timely manner allows for appropriate disease management and can significantly improve a patient's quality of life. Whole-exome sequencing (WES) is a robust and one of the most comprehensive genetic tests to identify the disease-causing changes in a large variety of genetic disorders.

In WES, protein-coding regions of all genes (~20,000) of the human genome, i.e. exome, are sequenced using next-generation sequencing technologies. While the exome constitutes of only ~1% of the whole genome, 85% of all disease-causing mutations are located there.

The diagnostic yield of WES outmatches those obtained with traditional gene diagnostic methods. A definite diagnosis is typically obtained in 20-60% of cases, depending on the medical specialty, with severe, early-onset disorders have the highest diagnostic rates.

WES is most suitable for individuals with

- a complex, unspecific genetic disorder with multiple differential diagnoses.
- a genetically highly heterogeneous disease.
- a suspected genetic disorder where a specific genetic test is not available.
- unsuccessful previous genetic testing.

Including parents or other family members further helps to obtain a correct genetic diagnosis. Performing WES for both the patient and parents is a necessity for direct detection of new mutations that are not present in parents but occur either in the formation of eggs or sperm cells or early in the development. These changes account for the majoity of severe developmental disorders.

For more information on genetic testing for patients and family members, please visit:
http://blueprintgenetics.com/what-we-do/for-patients/

I confirm that the information below has been explained to me concerning the test:

1. The results of this test may show that I and/or my family members have an inherited disease or are at an increased risk to be affected by a genetic disease. I understand that this test may detect previously unrecognized biological relationships, such as non-paternity.
2. I am aware that the results of this test might be inconclusive about my genetic status. While some genetic variants are known to be disease causing and others are known to be benign, a portion of genetic variants found are of uncertain significance. Depending on the results of this test, my physician may recommend genetic counseling or further testing of myself and/or my family members.
3. I understand that an anonymized summary of results from this test may be presented for example at meetings, scientific publications and/or DNA variant databases in order to improve the understanding, diagnostics and treatment of similar clinical conditions. No identifying information will ever be presented.
4. If I have selected the patient insurance billing option, I authorize my health plan or insurance provider to pay my insurance benefits directly to Blueprint Genetics. I authorize Blueprint Genetics to release information concerning my testing to my insurer. I understand that I am legally responsible for sending Blueprint Genetics any money received from my insurance company for performance of this genetic test. If my insurance does not cover these services or only covers part of the amount, I am responsible for remaining costs of this test.
5. I am aware that not consenting to any of the sections to follow will not in any way affect my further treatment. If no box is checked in a section, it is assumed that no consent is given.

6. **Separate consent for sample storage at Blueprint Genetics for 3 years for the purposes of family member testing.** By checking the relevant box below I give my consent to the 3-year storage of the DNA sample in the diagnostic laboratory of Blueprint Genetics for the purposes of family member testing. Without this permission the sample will be stored approximately for 12 months.

☐ I give my consent to the 3-year storage of the sample for family member testing.
☐ I do not give my consent to the 3-year storage of the sample family member testing.

7. **Separate consent for research use and long-term storage.** By checking the relevant box below I give my consent to the long-term storage of the DNA sample in the diagnostic laboratory of Blueprint Genetics (without separate consent for long-term storage the DNA samples are typically stored for approximately 12 months) for use of the DNA sample in research into hereditary Mendelian diseases and the efforts to improve the diagnostics and treatment of said diseases. The research data concerning me will be treated as confidential information and coded in such a way that my identity cannot be discovered without the key code in the possession of the Blueprint Genetics research physician. Where necessary, such coded research data may also be processed within or outside the European Union and released for use by another research group or a company participating in the study. I hereby give my consent to the use of the aforementioned research data for the purposes set out in this consent. The data will be preserved for 50 years.

   I understand that my consent to the research use of the sample taken for diagnostic purposes is voluntary and that I may cancel this consent and withdraw my participation at any time prior to the completion of the study. I am aware that the data collected up to the date of my withdrawal will be used as part of the research material.

☐ I give my consent to the research use and long-term storage of the sample as set out in Section 7 above.
☐ I do not give my consent to the research use and long-term storage of the sample as set out in Section 7 above.

8. **Separate consent for reporting of secondary findings.** By checking the relevant box below I give Blueprint Genetics my consent to report to my ordering healthcare professional any possible secondary findings that are not directly related to the reason for ordering my test. Blueprint Genetics reports as secondary findings pathogenic and likely pathogenic variants in selected genes associated with various genetic disorders. The selected genes where secondary findings are reported represent those included in "ACMG Recommendations for Reporting of Secondary Findings in Clinical Exome and Genome Sequencing" published by the American College of Medical Genetics and Genomics.

   I understand that secondary findings are of medical value and may have implications for my future health and for family planning purposes. I understand that the absence of secondary findings for any particular gene does not mean that there are no pathogenic variants in that gene.

   Blueprint Genetics needs to receive this consent within 28 days from sample reception in order to report any secondary findings. I understand that my family members can decide on their secondary findings independent of my decision.

☐ I give my consent to the reporting of secondary findings.
☐ I do not give my consent to the reporting of secondary findings.

☐ I give Blueprint Genetics permission to contact me regarding further genetic research and/or other genetic services relevant to me in the future. I may withdraw from such contact at any time.

PATIENT SIGNATURE

By signing this form, I acknowledge that I have read the Informed Consent for Whole Exome Sequencing and understand its content. I have had the opportunity to ask questions about this form and my questions have been answered.	
Name (please print):	**DOB:**
Signature:	**Date:**
Name and relationship of Legal Representative, if patient is a minor (please print):	**Signature of Legal Representative, if patient is a minor:**