Whole Exome Requisition Form

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

REQUIRED FIELDS ARE MARKED WITH AN ASTERISK (*)

Promotion/Contract Code: .....

TEST SELECTION*

☐ **Whole Exome** ☐ **Whole Exome Family**[i]	[i] Sample and Informed Consent is needed from all family members included in the Family test.
☐ **Expand to Exome**[ii] **Specify the previous order ID:**	[ii] Expand your patient's previous test done at Blueprint Genetics (panel, single gene) to Whole Exome or Whole Exome Family. You will be contacted if new sample is needed.

All tests include analysis of both small exonic and splice site variations, and large deletions and insertions.

Sample Type[iii]**:** ☐ **Blood** ☐ **Saliva** ☐ **DNA, source:**		**Sample Collection Date:**
Is the index patient or any of the family members a fetal sample? ☐	**Is this sample from an ongoing pregnancy?** ☐	**Gestational age:**
Maternal Cell Contamination (MCC) test done locally: ☐ **No** ☐ **Test ongoing** ☐ **Yes, no MCC identified** ☐ **Yes, MCC identified** Specify the level of MCC (%):		

[iii] Please note that this information affects interpretation for mitochondrial DNA testing. More information about sample requirements on blueprintgenetics.com/sample-requirements.

ORDERING HEALTHCARE PROFESSIONAL INFORMATION

***Name:**		***Institution:**
***Street Address:**		***Email:**
***City:**	**State:**	**Phone:**
***Zip/Post Code:**	***Country:**	**Fax:**
Delivery of results ☐ **Mail** ☐ **Fax** ☑ **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 A COLLEAGUE

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

PATIENT INFORMATION

To enable processing, provide at least two unique patient identifiers that match those on the sample label (we recommend using the patient's full name: first & last name and date of birth).

***First Name:**	***Last Name:**
***DOB:** Year / Month / Day	**Patient Identifier/MRN:**

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

First Name:*	**Last Name:***	**DOB:***	**Identifier /MRN:**
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:***	**Identifier /MRN:**
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:**
*** Has the patient or either of the family members received a hematopoietic stem cell transplantation?** ☐ Yes ☐ No	**Transplantation date:**
*** Has the patient or either of the family members received granulocyte transfusions in the past two weeks?** ☐ Yes ☐ No	**Transfusion date:**
Age of Primary Diagnosis:	
Has the patient died? ☐ Yes ☐ No **When?**	
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.	
Affected family members: ☐ Yes ☐ No **Who and what symptoms?**	
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

Please select **either** Institutional Billing **or** Patient Payment

☐ **INSTITUTIONAL BILLING**

Order reference(s):
Information added to this field will be visible on the invoice and medical report.

Purchase Order number:

Please provide all details below if ordering for the first time or billing different than usual institution for this order.

Facility Name:

Address:

City:	**State:**
Zip/Post Code:	**Country:**

Contact Person:

Email:

☐ **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 note that we accept Visa and Mastercard.

Please contact support@blueprintgenetics.com if you wish to discuss alternative payment options.

*** Email:**

*** Name:**

ORDERING HEALTHCARE 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 Blueprint Genetics' General Terms of Service, as currently posted at https://blueprintgenetics.com/general-terms/ . Unless there is a written agreement between the Institution and Blueprint Genetics, I accept, and have the authority to accept, these General Terms of Service on behalf of the Institution.	
*** Signature:**	
*** Name:**	*** Date (YYYY-MM-DD):**

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/

HEALTHCARE PROFESSIONAL SIGNATURE REQUIRED FOR PROCESSING

The Informed Consent for Whole Exome Sequencing is available in different languages 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

For more information on genetic testing for patients and family members, please visit: https://blueprintgenetics.com/resources/whole-exome-sequencing-guide-for-patients-and-families/

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 and it is disposed of after that unless earlier disposal is required by applicable laws.

   ☐ I give my consent to the 3-year storage of the sample for 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.

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 before sample is put into analysis 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.

More information about how we process personal data: https://blueprintgenetics.com/privacy/

☐ 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.	
Patient name (please print):	**Patient date of birth (YYYY-MM-DD):**
Patient signature:	**Date (YYYY-MM-DD):**
Name and relationship of Legal Representative, if patient is a minor (please print):	**Signature of Legal Representative, if patient is a minor:**