# 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 are also available.



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REQUIRED FIELDS ARE MARKED WITH AN ASTERISK (\*) Promotion/Contract Code: **TEST SELECTION\*** ^Sample and Informed Consent is needed from all family members ☐ Whole Exome Family Plus^ ■ Whole Exome Plus included in the Family test. All tests include analysis of both small exonic and splice site variations, and large deletions and insertions. Sample Type^^: Blood ■ Saliva ■ DNA, source: Sample Collection Date: ^^ 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** Fax V 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: Results can be shared within the same hospital on our ordering portal, Nucleus Phone: **Mail Results Nucleus** Fax: Fax Results **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) Identifier First Name:\* Last Name: DOB:\* /MRN: Phenotype Description: Relationship to Patient:\* Is Family Member 1 Affected With the Same Phenotype as Patient:\* ☐ No Partially Uncertain FAMILY MEMBER 2 INFORMATION (FILL ONLY IF WHOLE EXOME FAMILY PRODUCT IS ORDERED) Identifier First Name:\* Last Name:\* DOB:\* **Phenotype Description:** Relationship to Patient:\* Is Family Member 2 Affected With the Same Phenotype as Patient:\* Uncertain

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

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☐ No

Partially

# 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 Member	* Has the Patient or Either of the Family Members Received a Hematopoietic Stem Cell Transplantation?  Yes  No					
* Has the Patient or Either of the Family Members Received Granulocyte Transfusions in the Past Two Weeks?  Yes  No						
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 A	Abnormal Results:			
Please specify genes of interest:  Please specify suspected differential diagnosis (if applicable):		ea aitterential diagnosis (it applicable):				
CLINICAL FEATURES CHECKLIST						
Perinatal History	Endocrinological		Malformations – Brain			
☐ Cystic hygroma	☐ Diabetes mellitus		☐ Abnormalities of basal ganglia			
☐ Increased nuchal translucency	☐ Hyperparathyroidis		☐ Agenesis of the corpus callosum			
☐ Intrauterine growth restriction	☐ Hypoparathyroidisn	n	☐ Brain atrophy			
□ Oligohydramnios	☐ Hyperthyroidism		☐ Cortical dysplasia			
□ Polyhydramnios	☐ Hypothyroidism		☐ Hemimegalencephaly			
□ Prematurity	□ Paraganglioma		☐ Heterotropia			
☐ Other:	☐ Pheochromocytoma	a	☐ Holoprosencephaly			
	☐ Other:		☐ Hydrocephalus			
Cardiovascular			☐ Lissencephaly			
☐ Angioedema	Gastroenterological		☐ Macrocephaly			
☐ Aortic dilatation	☐ Constipation		☐ Microcephaly			
☐ Arrhythmia / conduction defect	☐ Chronic diarrhea		☐ Periventricular leukomalacia			
☐ Atrial septal defect	☐ Chronic intestinal p		☐ Other:			
☐ Cardiomyopathy	☐ Elevated transamina		Malfarmations Skalatel and Other			
☐ Coarctation of aorta	☐ Gastroesophageal r☐ Gastroschisis	епих	Malformations - Skeletal and Other			
<ul><li>☐ Hypoplastic left heart</li><li>☐ Malformation of heart and/or great</li></ul>	☐ Gastroschisis☐ Hepatic failure		☐ Cleft lip / palate ☐ Club foot / feet			
vessels	☐ Hirschsprung diseas	00	☐ Contractures			
□ Stroke	☐ Pyloric stenosis	JC	☐ Contractures ☐ Craniosynostosis			
☐ Tetralogy of Fallot	☐ Recurrent vomiting		☐ Dysmorphic features			
☐ Ventricular septal defect	☐ Tracheoesophageal	fistula	☐ Ear malformation			
☐ Other:	□ Other:		☐ Fractures ☐ Limb anomaly			
Dermatological	Hematological and Imm	nunological	☐ Overgrowth			
☐ Blistering	☐ Anemia		☐ Polydactyly			
☐ Connective tissue abnormality	☐ Coagulation disorde	er	☐ Scoliosis			
☐ Hair abnormality	☐ Immunodeficiency		☐ Short stature			
☐ Pigmentation abnormality	☐ Myelofibrosis		☐ Syndactyly			
☐ Ichthyosis	☐ Neutropenia		☐ Vertebral anomaly			
☐ Skin tumors	□ Pancytopenia		☐ Other:			
☐ Nail abnormality	☐ Thrombocytopenia					
☐ Other:	☐ Other:		1			

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# CLINICAL FEATURES CHECKLIST

CLINICAL FEATURES CHECKLIST		
Metabolic	Neuromuscular	Reproductive System Abnormalities
<ul> <li>Abnormal creatine phosphokinase</li> </ul>	☐ Ataxia	☐ Ambiguous genitalia
☐ Elevated alanine	☐ Chorea	☐ Cryptochidism
☐ Elevated puryvate	☐ Dystonia	☐ Hypogonadism
☐ Failure to thrive	☐ Hypotonia	☐ Hypospadias
☐ Ketosis	☐ Hypertonia	☐ Infertility
☐ Lactic acidosis	☐ Muscle weakness	☐ Undescended testis
☐ Organic aciduria	☐ Muscular dystrophy	☐ Other:
☐ Other:	☐ Neuropathy	Tumors / Malignancies
Nephrological	☐ Spasticity	☐ Adenomatous polyposis
☐ Hydronephrosis	☐ Other:	☐ Brain tumor
☐ Kidney malformation	Ophthalmological	☐ Breast cancer
☐ Renal agenesis or dysgenesis	☐ Abnormal eye movement	☐ Colorectal cancer
□ Renal tubulopathy	☐ Abnormal vision	☐ Leukemia
☐ Other:	☐ Blindness	☐ Lung cancer
Neurodevelopmental	☐ Cataracts	☐ Melanoma
□ ADHD	☐ Coloboma	☐ Other:
☐ Autism spectrum disorder	☐ CPEO	
Developmental delay	☐ Optic atrophy	
☐ Developmental regression	☐ Ptosis	
☐ Encephalopathy	□ Retinitis pigmentosa	
☐ Fine motor delay	☐ Other:	
☐ Gross motor delay		
☐ Hearing loss		
☐ Intellectual disability		
☐ Learning disability		
☐ Obsessive-compulsive disorder		
☐ Psychiatric symptoms		
☐ Recurrent headache		
☐ Seizures		
☐ Speech delay		
□ Other:		

# \* BILLING INFORMATION

LETTER

Please select either Insurance Billing or Institutional Billing or Patient Payment

	,			
INSURANCE BILLING		INSTITUTIONAL BILLING	PATIENT PAYMENT	
Include copy of both sides of the insurance card. The insurance provider usually requests a letter of medical necessity after submission of the claim.		Please provide all details below if ordering for the first time or billing different than usual institution for this order.  * ICD-10 Codes:	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	
* ICD-10 Codes:		Hospital Status:	sample goes to analysis once the payment has been collected by Blueprint Genetics.	
		Inpatient Outpatient Patient	Please note that we accept Visa and Mastercard.	
* Hospital Status Inpatient Outpatie  * Insurance Company:	ent Non-hospital patient	Facility Name:	Please contact billing.us@blueprintgenetics.com or (650) 452-9340 ext 1 if you wish to discuss alternative payment options.	
Insurance ID #:		Street Address:		
Group #:			* ICD-10 Codes:	
		City:		
Patient Relation to Policy H	older:		* Hospital Status:  Non-hospital	
Self Spouse	Child Other	State:	Inpatient Outpatient patient	
Policy Holder First Name:	Policy Holder Last Name:		* Email:	
		Zip/Post Code:		
Policy Holder DOB:				
<u> </u>		Country:	* First Name:	
Patient Street Address:				
		Contact Person:	* Last Name:	
Patient Address City:				
		Phone:		
Patient Address State:	Patient Address Zip Code:	<u> </u>	Phone:	
		Fax:		

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## **ORDERING HEALTHCARE PROFESSIONAL SIGNATURE**

To ensure compliance with state law, verification of patient informed consent is required for genetic testing. Testing laboratories located in Massachusetts require a signed acknowledgment from the ordering medical practitioner. This signed acknowledgment is required to complete the genetic testing you've ordered: I acknowledge that prior to ordering genetic testing on the patient listed above, I have obtained a signed, written consent form from the patient (or their authorized representative) and possible family members included in the test (Whole Exome Family products), as required by applicable state law and/or regulations. Additionally, I will maintain all written consent forms as part of the patient file and will make them available to Blueprint Genetics upon reasonable request.

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.

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/

#### HEALTH CARE 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.
- 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.	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.					
Мо	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.  ATIENT SIGNATURE					
By signir opportu	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 (pl	ease print):	Date of birth (YYYY-MM-DD):				
Signature	::	Date (YYYY-MM-DD):				
Name and	d relationship of Legal Representative, if patient is a minor (please print):	Signature of Legal Representative, if patient is a minor:				

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