

# Whole Exome Requisition Form

This requisition form, and consent forms in other languages, can be printed from [www.blueprintgenetics.com](http://www.blueprintgenetics.com), where test codes and detailed descriptions on tests are also available.

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Promotion/Contract Code: ..... CLIA# 99D2092375, CAP# 9257331

REQUIRED FIELDS ARE MARKED WITH AN ASTERISK (\*)

## TEST SELECTION\*

<input type="checkbox"/> Whole Exome Plus	<input type="checkbox"/> Whole Exome Family Plus^	^Sample and Informed Consent is needed from all family members included in the Family test.
<input type="checkbox"/> 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.

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

Sample Type^^:	<input type="checkbox"/> Blood	<input type="checkbox"/> Saliva	<input type="checkbox"/> DNA, source:	Sample Collection Date:
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^^ Please note that this information affects interpretation for mitochondrial DNA testing. More information about sample requirements on [blueprintgenetics.com/sample-requirements](http://blueprintgenetics.com/sample-requirements).

## ORDERING HEALTH CARE PROFESSIONAL INFORMATION

Name and Full Address:*	Institution:*		
	Email:*		
	NPI# (REQUIRED, US only):		
	Phone:		
	Fax:		
<input type="checkbox"/> Mail Results	<input type="checkbox"/> Fax Results	<input checked="" type="checkbox"/> Nucleus	Results will always be available on our online reporting system at <a href="http://nucleus.blueprintgenetics.com">nucleus.blueprintgenetics.com</a>

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:	<input type="checkbox"/> Fax Results	<input type="checkbox"/> Mail Results
		<input type="checkbox"/> 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:*			
<input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Partially <input type="checkbox"/> 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:*			
<input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Partially <input type="checkbox"/> 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)

<b>Sex:*</b> <input type="checkbox"/> Male <input type="checkbox"/> Female <input type="checkbox"/> Unknown/uncertain		<b>Ethnicity:</b>	
<b>ICD-10 Codes:*</b> (required for patient insurance – US only)			
<b>Has the Patient Died?</b> <input type="checkbox"/> Yes <input type="checkbox"/> No		<b>Hospital Status:</b> <input type="checkbox"/> Inpatient <input type="checkbox"/> Outpatient <input type="checkbox"/> Non-hospital patient	
<b>Has the Patient or Either of the Family Members Received a Hematopoietic Stem Cell Transplantation?*</b> <input type="checkbox"/> No <input type="checkbox"/> Yes, specify who:		<b>Has the Patient or Either of the Family Members Received Granulocyte Transfusions in the Past Two Weeks?*</b> <input type="checkbox"/> No <input type="checkbox"/> Yes, specify who:	
<b>Describe All Clinical Findings *</b> (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.			
<b>Family History</b> (e.g. parental consanguinity, attach pedigree if available):			
<b>Previous Testing With Normal Results:</b>		<b>Previous Testing With Abnormal Results:</b>	
<b>Please specify genes of interest:</b>		<b>Please specify suspected differential diagnosis (if applicable):</b>	

**CLINICAL FEATURES CHECKLIST**

<p><b>Perinatal History</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Cystic hygroma</li> <li><input type="checkbox"/> Increased nuchal translucency</li> <li><input type="checkbox"/> Intrauterine growth restriction</li> <li><input type="checkbox"/> Oligohydramnios</li> <li><input type="checkbox"/> Polyhydramnios</li> <li><input type="checkbox"/> Prematurity</li> <li><input type="checkbox"/> Other:.....</li> </ul> <p><b>Cardiovascular</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Angioedema</li> <li><input type="checkbox"/> Aortic dilatation</li> <li><input type="checkbox"/> Arrhythmia / conduction defect</li> <li><input type="checkbox"/> Atrial septal defect</li> <li><input type="checkbox"/> Cardiomyopathy</li> <li><input type="checkbox"/> Coarctation of aorta</li> <li><input type="checkbox"/> Hypoplastic left heart</li> <li><input type="checkbox"/> Malformation of heart and/or great vessels</li> <li><input type="checkbox"/> Stroke</li> <li><input type="checkbox"/> Tetralogy of Fallot</li> <li><input type="checkbox"/> Ventricular septal defect</li> <li><input type="checkbox"/> Other:.....</li> </ul> <p><b>Dermatological</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Blistering</li> <li><input type="checkbox"/> Connective tissue abnormality</li> <li><input type="checkbox"/> Hair abnormality</li> <li><input type="checkbox"/> Pigmentation abnormality</li> <li><input type="checkbox"/> Ichthyosis</li> <li><input type="checkbox"/> Skin tumors</li> <li><input type="checkbox"/> Nail abnormality</li> <li><input type="checkbox"/> Other:.....</li> </ul>	<p><b>Endocrinological</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Diabetes mellitus</li> <li><input type="checkbox"/> Hyperparathyroidism</li> <li><input type="checkbox"/> Hypoparathyroidism</li> <li><input type="checkbox"/> Hyperthyroidism</li> <li><input type="checkbox"/> Hypothyroidism</li> <li><input type="checkbox"/> Paraganglioma</li> <li><input type="checkbox"/> Pheochromocytoma</li> <li><input type="checkbox"/> Other:.....</li> </ul> <p><b>Gastroenterological</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Constipation</li> <li><input type="checkbox"/> Chronic diarrhea</li> <li><input type="checkbox"/> Chronic intestinal pseudo-obstruction</li> <li><input type="checkbox"/> Elevated transaminases</li> <li><input type="checkbox"/> Gastroesophageal reflux</li> <li><input type="checkbox"/> Gastroschisis</li> <li><input type="checkbox"/> Hepatic failure</li> <li><input type="checkbox"/> Hirschsprung disease</li> <li><input type="checkbox"/> Pyloric stenosis</li> <li><input type="checkbox"/> Recurrent vomiting</li> <li><input type="checkbox"/> Tracheoesophageal fistula</li> <li><input type="checkbox"/> Other:.....</li> </ul> <p><b>Hematological and Immunological</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Anemia</li> <li><input type="checkbox"/> Coagulation disorder</li> <li><input type="checkbox"/> Immunodeficiency</li> <li><input type="checkbox"/> Myelofibrosis</li> <li><input type="checkbox"/> Neutropenia</li> <li><input type="checkbox"/> Pancytopenia</li> <li><input type="checkbox"/> Thrombocytopenia</li> <li><input type="checkbox"/> Other:.....</li> </ul>	<p><b>Malformations – Brain</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Abnormalities of basal ganglia</li> <li><input type="checkbox"/> Agenesis of the corpus callosum</li> <li><input type="checkbox"/> Brain atrophy</li> <li><input type="checkbox"/> Cortical dysplasia</li> <li><input type="checkbox"/> Hemimegalencephaly</li> <li><input type="checkbox"/> Heterotropia</li> <li><input type="checkbox"/> Holoprosencephaly</li> <li><input type="checkbox"/> Hydrocephalus</li> <li><input type="checkbox"/> Lissencephaly</li> <li><input type="checkbox"/> Macrocephaly</li> <li><input type="checkbox"/> Microcephaly</li> <li><input type="checkbox"/> Periventricular leukomalacia</li> <li><input type="checkbox"/> Other:.....</li> </ul> <p><b>Malformations - Skeletal and Other</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Cleft lip / palate</li> <li><input type="checkbox"/> Club foot / feet</li> <li><input type="checkbox"/> Contractures</li> <li><input type="checkbox"/> Craniosynostosis</li> <li><input type="checkbox"/> Dysmorphic features</li> <li><input type="checkbox"/> Ear malformation</li> <li><input type="checkbox"/> Fractures</li> <li><input type="checkbox"/> Limb anomaly</li> <li><input type="checkbox"/> Overgrowth</li> <li><input type="checkbox"/> Polydactyly</li> <li><input type="checkbox"/> Scoliosis</li> <li><input type="checkbox"/> Short stature</li> <li><input type="checkbox"/> Syndactyly</li> <li><input type="checkbox"/> Vertebral anomaly</li> <li><input type="checkbox"/> Other:.....</li> </ul>
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**CLINICAL FEATURES CHECKLIST**

<p><b>Metabolic</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Abnormal creatine phosphokinase</li> <li><input type="checkbox"/> Elevated alanine</li> <li><input type="checkbox"/> Elevated puruvate</li> <li><input type="checkbox"/> Failure to thrive</li> <li><input type="checkbox"/> Ketosis</li> <li><input type="checkbox"/> Lactic acidosis</li> <li><input type="checkbox"/> Organic aciduria</li> <li><input type="checkbox"/> Other:.....</li> </ul> <p><b>Nephrological</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Hydronephrosis</li> <li><input type="checkbox"/> Kidney malformation</li> <li><input type="checkbox"/> Renal agenesis or dysgenesis</li> <li><input type="checkbox"/> Renal tubulopathy</li> <li><input type="checkbox"/> Other:.....</li> </ul> <p><b>Neurodevelopmental</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> ADHD</li> <li><input type="checkbox"/> Autism spectrum disorder</li> <li><input type="checkbox"/> Developmental delay</li> <li><input type="checkbox"/> Developmental regression</li> <li><input type="checkbox"/> Encephalopathy</li> <li><input type="checkbox"/> Fine motor delay</li> <li><input type="checkbox"/> Gross motor delay</li> <li><input type="checkbox"/> Hearing loss</li> <li><input type="checkbox"/> Intellectual disability</li> <li><input type="checkbox"/> Learning disability</li> <li><input type="checkbox"/> Obsessive-compulsive disorder</li> <li><input type="checkbox"/> Psychiatric symptoms</li> <li><input type="checkbox"/> Recurrent headache</li> <li><input type="checkbox"/> Seizures</li> <li><input type="checkbox"/> Speech delay</li> <li><input type="checkbox"/> Other:.....</li> </ul>	<p><b>Neuromuscular</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Ataxia</li> <li><input type="checkbox"/> Chorea</li> <li><input type="checkbox"/> Dystonia</li> <li><input type="checkbox"/> Hypotonia</li> <li><input type="checkbox"/> Hypertonia</li> <li><input type="checkbox"/> Muscle weakness</li> <li><input type="checkbox"/> Muscular dystrophy</li> <li><input type="checkbox"/> Neuropathy</li> <li><input type="checkbox"/> Spasticity</li> <li><input type="checkbox"/> Other:.....</li> </ul> <p><b>Ophthalmological</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Abnormal eye movement</li> <li><input type="checkbox"/> Abnormal vision</li> <li><input type="checkbox"/> Blindness</li> <li><input type="checkbox"/> Cataracts</li> <li><input type="checkbox"/> Coloboma</li> <li><input type="checkbox"/> CPEO</li> <li><input type="checkbox"/> Optic atrophy</li> <li><input type="checkbox"/> Ptosis</li> <li><input type="checkbox"/> Retinitis pigmentosa</li> <li><input type="checkbox"/> Other:.....</li> </ul>	<p><b>Reproductive System Abnormalities</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Ambiguous genitalia</li> <li><input type="checkbox"/> Cryptochidism</li> <li><input type="checkbox"/> Hypogonadism</li> <li><input type="checkbox"/> Hypospadias</li> <li><input type="checkbox"/> Infertility</li> <li><input type="checkbox"/> Undescended testis</li> <li><input type="checkbox"/> Other:.....</li> </ul> <p><b>Tumors / Malignancies</b></p> <ul style="list-style-type: none"> <li><input type="checkbox"/> Adenomatous polyposis</li> <li><input type="checkbox"/> Brain tumor</li> <li><input type="checkbox"/> Breast cancer</li> <li><input type="checkbox"/> Colorectal cancer</li> <li><input type="checkbox"/> Leukemia</li> <li><input type="checkbox"/> Lung cancer</li> <li><input type="checkbox"/> Melanoma</li> <li><input type="checkbox"/> Other:.....</li> </ul>
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**BILLING INFORMATION\***

<input type="checkbox"/> <b>INSURANCE BILLING</b> Include copy of both sides of the insurance card. The insurance provider usually requests a letter of medical necessity after submission of the claim.	<input type="checkbox"/> <b>INSTITUTIONAL BILLING</b> Facility Name:	<input type="checkbox"/> <b>PATIENT PAYMENT</b> 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 <a href="mailto:billing.us@blueprintgenetics.com">billing.us@blueprintgenetics.com</a> or (650) 452-9340 ext 1 if you wish to discuss alternative payment options.
Insurance Company:	Street Address:	
Insurance ID #:		
Group #:	City:	State:
Patient Relation to Policy Holder: <input type="checkbox"/> Self <input type="checkbox"/> Spouse <input type="checkbox"/> Child <input type="checkbox"/> Other	Zip/Post Code:	Country:
Policy Holder Name:	Contact Person:	Name:
Policy Holder DOB:	Phone:	Fax:
		Phone:

<p><b>ORDERING HEALTH CARE PROFESSIONAL SIGNATURE</b></p>	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/](http://www.blueprintgenetics.com/how-to-order/). **When ordering a Whole Exome Family product, please print out a separate Informed Consent for each family member.**

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## 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 majority 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.

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- 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 DOB:
Patient signature:	Date:
Name and relationship of Legal Representative, if patient is a minor (please print):	Signature of Legal Representative, if patient is a minor: