

Raw Sequence Data Consent Form

Proband Name : _____ DOB : _____

Ambyr Genetics provides the raw sequence data when requested, with the understanding that the data will be used strictly on a research basis, and not for clinical purposes. Raw sequence data refers to the unanalyzed sequence data produced from sequencing of the proband and additional family members. Specialized computer software is required to view and interpret this data. In the case that additional important findings related to the phenotype in question are identified, clinicians should immediately contact the laboratory for verification, and possibly, generation of an amended report. All patients undergoing diagnostic exome sequencing (DES) have completed a consent form which includes the opportunity to opt-out of secondary findings disclosure.

Note: Raw data are not released until clinical testing is completed and a final report has been issued.

Raw Sequence Data Options (please choose one):

Downloadable from ftp server (no charge):

- fastq files
 BAM files
 VCF files

Shipped on hard drive (\$100):

- fastq files BAM files
 Shipping costs (ground shipping) (\$50) or provide courier account number: _____

The requesting physician and all authorized recipients will be notified by e-mail when the raw data is available for download or has been shipped, usually 4-6 weeks from the final report date.

TOTAL \$: _____

Credit card # : _____

Name on card: _____ Exp. date : _____

Signature : _____

PATIENT/GUARDIAN CONSENT

I understand that my doctor has requested receipt of the raw sequence data resulting from the diagnostic exome sequencing (DES) performed for me/the person for whom I am the caregiver. I acknowledge that the information included in the data files may include secondary findings, potential sequencing artifacts, and variants which have not undergone interpretation. I also understand that these data are for research purposes only and shall not be used for clinical decision-making purposes. I understand that Ambyr Genetics recommends against the delivery of these data to patients.

NAME AND DOB OF EACH PATIENT/FAMILY MEMBER FROM WHOM YOU ARE REQUESTING RAW SEQUENCE DATA:

NAME	DOB	PATIENT/GUARDIAN SIGNATURE	DATE
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.....
.....

PHYSICIAN CONSENT

I understand that the receipt of raw sequence data may include secondary findings, potential sequencing artifacts, and variants of which have not undergone interpretation and the patient/family members listed above are aware I am requesting this data. I also understand that any information gleaned from review of this data, outside that of which is described in the patient's final report, is strictly for research purposes and shall not be used for clinical decision-making purposes. I understand that Ambyr Genetics recommends against the delivery of these data to patients.

Signature : _____

Date : _____

Printed Name : _____

Phone : _____

Institution : _____

Email Address : _____

AUTHORIZED DATA RECIPIENTS

Raw exome data can be made available to the following individuals by request. Each authorized recipient may contact the laboratory to request raw data for the patient.

Name : _____

Email : _____

Name : _____

Email : _____