

# Critical WES Trio

by **Baylor Miraca Genetics Laboratories**

<b>Critical Whole Exome Sequencing Trio (Test Code 1722)</b>	
Test Information:	Whole Exome Sequencing Trio (WES trio) test is used when a patient's medical history and physical exam findings strongly suggest that there is an underlying genetic etiology. In some cases, the patient may have had an extensive evaluation consisting of multiple genetic tests, without identifying an etiology. In other cases, a physician may opt to order the Whole Exome Sequencing Trio test early in the patient's evaluation in an effort to expedite a possible diagnosis. This testing must be ordered by a physician and must be accompanied with a consent form and detailed clinical information.
Indications for Testing:	Patient with undiagnosed genetic syndrome and needs rapid turn-around time for results. This is recommended for patients who are critically ill. Parental samples are required to interpret WES trio results. See below and requisition for sample requirements and further details. Testing cannot be requested unless both parents submit samples for testing.
<b>Technical Information</b>	
Methodology:	Exome Capture and Next Generation Sequencing
<b>Sample &amp; Shipping Information</b>	
Specimen Type:	<b>Blood</b>
Requirements:	Draw blood in an EDTA (purple-top) tube(s) for the patient and both parents. Send at least 5cc (children) or 10cc (adults).
Specimen Type:	<b>Cultured Skin Fibroblast</b>
Requirements:	Send 2 T25 flasks at 80-100% confluence
Specimen Type:	<b>Purified DNA</b>
Requirements:	Send at least 20ug of purified DNA (minimal concentration of 50ng/uL; A260/A280 of ~1.7) for the patient and both parents.
Shipping Conditions:	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze. Sample must arrive within 48 hrs.
Turn Around Time:	Within 3 weeks
<b>Billing Information</b>	
List Price:	*For Prices, please call. Only institutional or self pay is acceptable for this test. We will not bill the patients insurance. This is to ensure that the rapid turnaround time can be met.