Ordering MBS funded Whole Exome Sequencing

Before ordering tests:

1. The requesting clinician must be a clinical geneticist or specialist paediatrician at RCH and have completed the RCH Learning Hero Genomics module.
2. Specialist paediatricians must discuss the case with a clinical geneticist, who will provide a test approval number if the patient is eligible. The test approval number must be provided in the order.
3. Confirm that the patient is an eligible out-patient and meets the definitions for Medicare Benefits Schedule Item number 73358 or 73359. As of July 2020 this definition is a patient aged 10 years or younger, strongly suspected of having a monogenic condition based on the presence of:

   (i) dysmorphic facial appearance and one or more major structural congenital anomalies; or

   (ii) intellectual disability or global developmental delay of at least moderate severity, as determined by a specialist paediatrician;

4. If a trio exome is required, parents must be registered so that an EMR record is created and testing can be ordered via EPIC.
5. If working on site, and family is present, ask the parents to get registered at the check-in desk

   If working off site, or family is not present, register parents by contacting HIS by telephone (urgent requests only) on 93456174 or by email His.patientreg@rch.org.au

   Information required:

   - Biological Mother and Father Surname and First name
   - Date of Birth
   - Sex
   - Address and telephone details
   - Medicare Number

6. Create a new documentation encounter in EPIC in the child's record and complete comprehensive phenotype information by using the smart phrase .exome

   The smart phrase guides the provision of clinical and phenotype information to assist in determining;

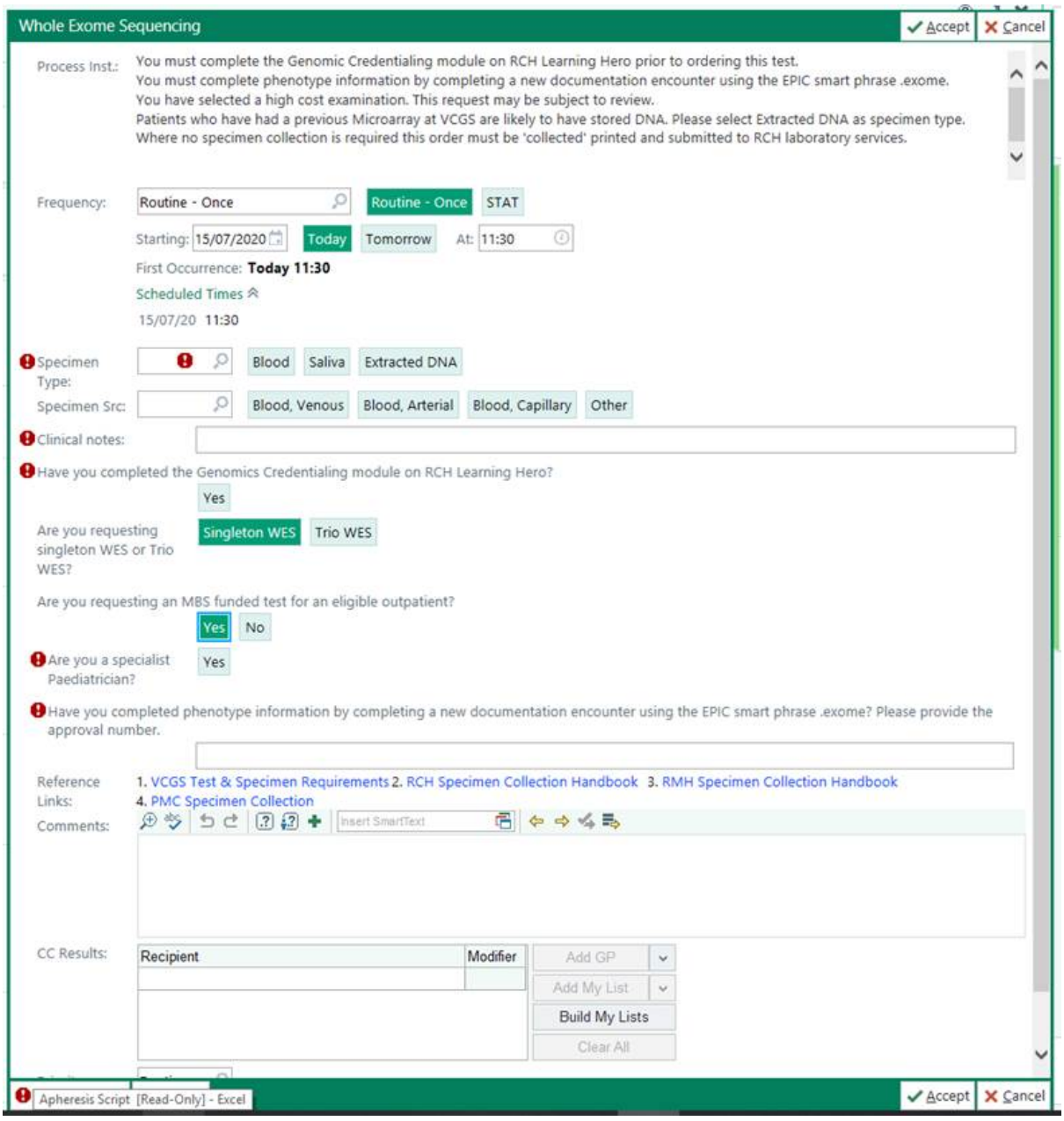
   - eligibility for MBS funding,
   - development of an appropriate phenotype-driven gene list to maximise test yield
   - The name of the clinical geneticist involved in the discussion and their approval number.

7. Share the documentation encounter by using the “send chart” function as an “e-communication” to the “genetics referral triage pool”

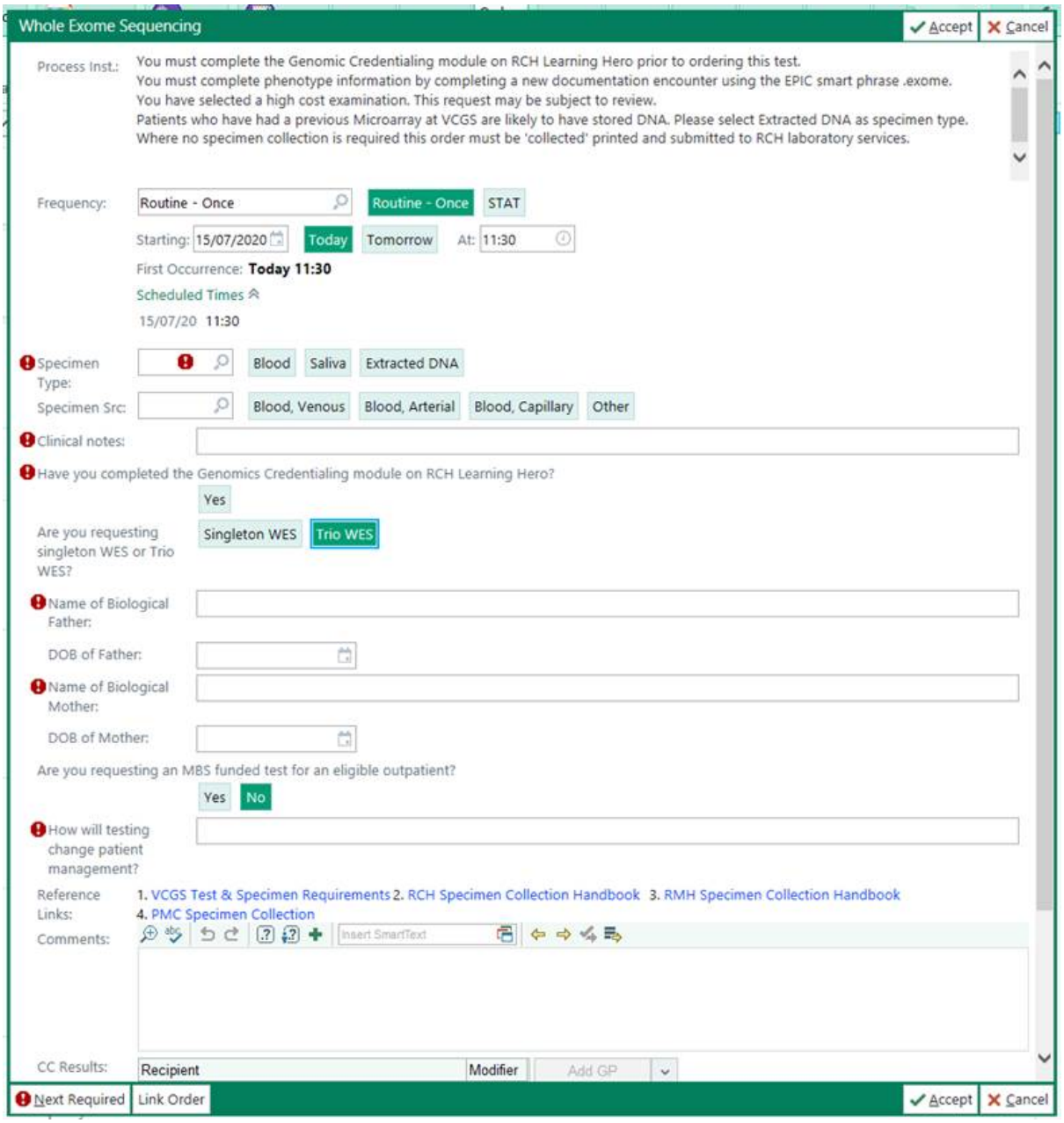
   Ordering the test

   Once approval has been obtained from a geneticist, you can order the test.

   In the child’s EMR place an order for
   - Whole exome sequencing (synonym, clinical exome)
   - Select singleton or Trio
   - The singleton request is show below

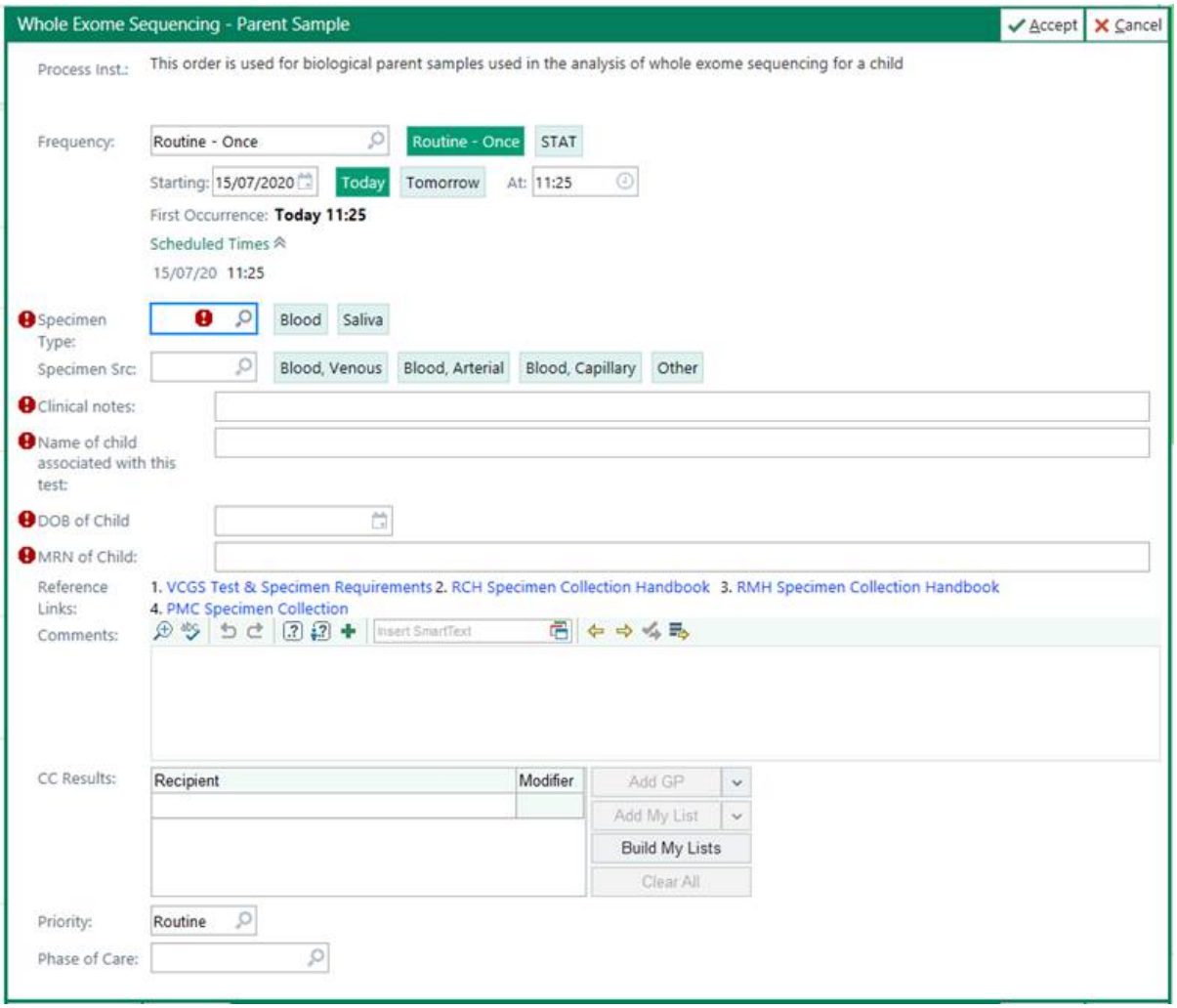


For Trio request, enter biological parent details for linking purposes



For Trio requests, request the biological parent orders within the EMR record of each parent

The order name is Whole exome sequencing-Parent sample


Whole Exome Sequencing - Parent Sample
Accept
Cancel
Process Inst.:
This order is used for biological parent samples used in the analysis of whole exome sequencing for a child
Frequency:
Routine - Once
Routine - Once
STAT
Starting:
15/07/2020
Today
Tomorrow
At:
11:25
First Occurrence: Today 11:25
Scheduled Times
15/07/20 11:25
Specimen Type:
Blood
Saliva
Specimen Src:
Blood, Venous
Blood, Arterial
Blood, Capillary
Other
Clinical notes:
Name of child associated with this test:
DOB of Child
MRN of Child:
Reference Links:
1. VCGS Test & Specimen Requirements 2. RCH Specimen Collection Handbook 3. RMH Specimen Collection Handbook
4. PMC Specimen Collection
Comments:
Insert SmartText
CC Results:
Recipient
Modifier
Add GP
Add My List
Build My Lists
Clear All
Priority:
Routine
Phase of Care: