ALBERTA PRECISION LABORATORIES
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Laboratory Bulletin

DATE:	9 December 2024
TO:	All Zones
FROM:	Genetics and Genomics, Molecular Genetics South, APL
RE:	**Updated Methylation-Specific MLPA (MS-MLPA) Kit for Russell-Silver Syndrome (RSS)**

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Key Message

- Previously, Molecular Genetics South used two different MS-MLPA kits to confirm a diagnosis of RSS
  - ME030-C3: chromosome 11
  - ME032-B1: chromosomes 7 and 14
- As of July 17, 2024, MRC Holland has replaced the ME030-C3 kit with the new ME030-D1 kit which now includes chromosomes 7 and 11.
- Molecular Genetics South will use the new ME030-D1 kit for RSS testing; the ME030-C3 kit will no longer be used for this indication.

Background

- RSS can be caused by abnormal methylation of chromosome 11p15 or maternal uniparental disomy (UPD) of chromosome 7. Molecular Genetics South offers testing for RSS by MS-MLPA, which can detect abnormal methylation of the two imprinted domains located within the chromosome 11p15 BWS/RSS gene cluster, abnormal methylation at 7p14-p11.1 and 7q32, as well as deletions and duplications within these regions. MS-MLPA will also detect UPD11 and UPD7 or infer an imprinting defect on chromosome 11 or 7.
- Previously, Molecular Genetics South used two different MS-MLPA kits to perform this testing. ME030-C3 included probes for the imprinted regions on chromosome 11, and ME032-B1 included probes for the imprinted regions on chromosomes 7 and 14. Therefore, patients who were undergoing testing for RSS were also having testing for RSS-like due to Temple Syndrome (UPD14mat).

How this will impact you

- Molecular Genetics South has validated the updated ME030-D1 kit and will use this kit for RSS testing moving forward. Patients undergoing testing for RSS will be assessed for abnormal methylation of chromosome 11p15 and UPD7. These patients will no longer have testing for UPD14 unless this is specifically requested.

Action Required

- Ordering physicians should continue to use the EPIC order, “Russell Silver Syndrome” (LAB4126) for RSS testing. If the ordering physician is querying RSS-like due to Temple Syndrome (UPD14mat), submit a separate Epic order for “Uniparental Disomy” (LAB4131) and write “chromosome 14” for the “specify chromosome” order question. Testing for RSS or UPD14 can also be requested using the Molecular Genetics Laboratory General Requisition (https://www.albertahealthservices.ca/frm-21892.pdf)

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Effective Immediately

Questions/Concerns

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Approved by

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