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Laboratory Bulletin

DATE:	23 December 2024
TO:	All Zones
FROM:	APL Genetics and Genomics, Molecular Genetics South
RE:	**Specimen Requirements for Beckwith-Wiedemann Syndrome and Russell Silver Syndrome**

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Key Message

- The preferred specimen types for Beckwith-Wiedemann Syndrome (BWS) & Russell Silver Syndrome (RSS) testing are whole blood, amniotic fluid, and embryonic fetal tissue.
- The Molecular Genetics South Lab (MGL) previously accepted extraembryonic tissue (umbilical cord or chorionic villi) if this was the only specimen type available.
- Effective immediately, MGL will no longer attempt BWS or RSS testing on extraembryonic tissue given that this is an indirect specimen type and may not represent the fetal methylation profile.

Background

- The majority of BWS and RSS cases are caused by abnormal methylation of the two imprinted domains located within the chromosome 11p15 BWS/RSS gene cluster, and more rarely by deletions and duplications within this region. RSS can also be caused by abnormal methylation at 7p14-p11.1 and 7q32 due to maternal uniparental disomy.
- MGL performs methylation-specific MLPA to detect abnormal methylation and copy number variation to provide a diagnosis of BWS or RSS. There is insufficient information available to suggest that abnormal methylation can be detected in extraembryonic tissues, given that the methylation status may not represent the fetal methylation profile.

How this will impact you

- BWS or RSS testing can be requested for whole blood, amniotic fluid or embryonic fetal tissue.

Action Required

- For prenatal testing, request BWS or RSS testing on amniotic fluid. For postnatal fetal testing, do not request testing on extraembryonic tissue specimens. Instead, obtain consent from the patient to test embryonic fetal tissue (ex. skin, muscle, etc.) which can be coordinated at the time of fetal autopsy.

Effective Immediately

Questions/Concerns

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Approved by

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