

| | | | | |
|---|---|------------------|--------------------------|------------------------------|
| Patient Name SAMPLEREP,SBMA A | Patient ID SA00058857 | Age 46 | Gender F | Order # SA00058857 |
| Ordering Phys CLIENT,CLIENT | | | DOB 06/10/1966 | |
| Client Order # SA00058857 | Account Information | | | Report Notes |
| Collected 06/06/2013 00:00 | C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901 | | | |
| Printed 06/11/2013 16:10 | | | | |

| Test | Flag | Results | Unit | Reference Value | Perform Site* |
|--|------|---|------|-----------------|---------------|
| Spinobulbar Musc Atrophy, Kennedy's | | | | | |
| RECEIVED: 06/07/2013 15:57 REPORTED: 06/10/2013 16:47 | | | | | |
| Specimen | | Blood | | | MCR |
| Specimen ID | | 1062124 | | | MCR |
| Order Date | | 10 Jun 2013 09:16 | | | MCR |
| Reason For Referral | | Test for the presence of an expansion within the androgen receptor gene. | | | MCR |
| Method | | A PCR-based assay was utilized to detect CAG-repeat expansions within the androgen receptor gene. We estimate that the number of CAG repeats is correct to within +/-5%. Normal: 11-34; indeterminate: 35-37; Full penetrance: 38-62. | | | MCR |
| Result | | CAG repeat: 22 (Normal) and 44 (Full penetrance) | | | MCR |
| Interpretation | | This result is consistent with this individual being a carrier of SBMA. | | | MCR |
| | | Since an expansion has been identified within the androgen receptor gene, testing of other at risk family members is possible. | | | |
| | | A genetic consultation may be of benefit. | | | |
| | | CAUTIONS: Test results should be interpreted in context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete. | | | |
| | | Rare polymorphisms exist that could lead to false negative or positive results. If results obtained do not match the clinical findings, additional testing should be considered. | | | |
| | | Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant. | | | |
| | | Laboratory developed test. | | | |
| Reviewed By: | | Emily Christine Lauer | | | MCR |
| Release Date | | 10 Jun 2013 16:45 | | | MCR |

Performing Site Legend on Last Page of Report

| | | |
|---|---|-------------------------------|
| Patient Name SAMPLEREP,SBMA A | Collection Date and Time 06/06/2013 00:00 | Report Status Final |
| Page 1 of 2 | >> Continued on Next Page >> | |

* Report times for Mayo performed tests are CST/CDT

| | | | | |
|---|---|------------------|--------------------|------------------------------|
| Patient Name SAMPLEREP,SBMA A | Patient ID SA00058857 | Age 46 | Gender F | Order # SA00058857 |
| Ordering Phys CLIENT,CLIENT | | | | DOB 06/10/1966 |
| Client Order # SA00058857 | Account Information | | | Report Notes |
| Collected 06/06/2013 00:00 | C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901 | | | |
| Printed 06/11/2013 16:10 | | | | |

| Test | Flag | Results | Unit | Reference Value | Perform Site* |
|------|------|---------|------|-----------------|---------------|
|------|------|---------|------|-----------------|---------------|

>> Accession SA00058857 - Continued From Previous Page <<
>> Do Not Discard <<

* Performing Site:

| | | |
|-----|---|---------------|
| MCR | Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905 | Lab Director: |
|-----|---|---------------|

| | | |
|---|---|-------------------------------|
| Patient Name SAMPLEREP,SBMA A | Collection Date and Time 06/06/2013 00:00 | Report Status Final |
| Page 2 of 2 | ** End of Report ** | |

* Report times for Mayo performed tests are CST/CDT