

WESSEX GENOMICS LABORATORY SERVICE (Salisbury) – RARE DISEASE TESTS – SERVICE LIST – fr ! "# A\$ril %&%'

N #Q NHS referrals in England are funded directly by NHS England and are subject to the National Genomic Test Directory; referral criteria must satisfy the NHSE Rare and Inherited Disease eligibility criteria. Test Directory indications are in b l*+ Tests not currently co ered by the Test Directory are mar!ed " ith an asteris!#. Non\$NHSE%&ri ate and

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<p>1ystic fibrosis Routine diagnostic or carrier testing R"38+"2 R"39+" =rgent carrier testing R"39+" Ne" born screen from blood s&ots R%9'</p>	<p>. / 8. . " or!ing days</p>	<p>C+TR C+TR C+TR</p>	<p>B> most common =6 mutations B> most common =6 mutations . most common =6 mutations</p>	<p>Targeted mutation test</p>
<p>'actor 2 *eiden and 3rothrombin common ariants# 'amilial testing for !no" n ariants 3redicti e testing R%8% 1arrier testing R%88 3arental0segregation testing R' 69</p>	<p>. / 8. . / . /</p>	<p>+5, +2 #lease noo</p>	<p>NGS genoty&ing</p>	<p>Targeted mutation test</p>

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<p>Sil er\$Russell syndrome 4Gro" th failure in early childhood5 R"86+%</p> <p>1 chromosome 88 abnormalities 411R8 only5 and =3D4F5 analysis</p> <p>=3D4885 follo" \$u& R%4'</p>	<p>./</p> <p>./</p>	<p>, %-.) +2(IG\$D, R% R6%9(alt\$TSS\$D, R% 7' "T(alt\$TSS\$D, R</p> <p>No s&ecific gene</p>	<p>, S\$, *3-</p> <p>, icrosatellite analysis</p>	<p>Im&rinting analysis</p>
<p>Thoracic aortic aneurysm or dissection 4T --D5 R""9</p>	<p>:.:</p>	<p>78\$gene &anel</p>	<p>Ne; t\$generation se<uencing ? , *3- for +6! % only</p>	<p>NGS ser ice</p>