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

N #() 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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|---|---|---|---|---|---|
| **1**ystic fibrosis<br>Routine diagnostic or carrier testing R**"38+"2**<br>R**"39+"**<br><br>**=**rgent carrier testing R**"39+"**<br><br>Ne**"** born screen from blood s**&**ots R**%9'** | | **. /**<br><br>**8.**<br><br>**. "**or**!**ing days | C**+**TR<br><br>C**+**TR<br><br>C**+**TR | **B>** most common **=6** mutations<br><br>**B>** most common **=6** mutations<br><br>**.** most common **=6** mutations | Targeted mutation test |
| **'**actor **2 ***eiden and **3**rothrombin common ariants**#** | | **. /** | **+5**, +2 | NGS genoty**&**ing | Targeted mutation test |
| **'**amilial testing for **!**no**"**n ariants<br>**3**redicti e testing R**%8%**<br><br>**1**arrier testing R**%88**<br><br>**3**arental**0**segregation testing R**'69** | | **8.**<br><br>**. /**<br><br>**. /** | **#**lease noo | | |

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|---|---|---|---|---|---|
| Sil er$Russell syndrome 4Gro " th failure in early childhood5 R"86+%<br>1hromosome 88 abnormalities 4l1R8 only5 and =3D4F5 analysis | | . / | , %-.) +2(IG$D , R%<br>R6%9(alt$TSS$D , R%<br>7 '"T(alt$TSS$D , R | , S$ , *3- | Im&rinting analysis |
| =3D488 5 follo " $u& R%4 ' | | . / | No s&ecific gene | , icrosatellite analysis | |
| Thoracic aortic aneurysm or dissection 4T- -D5 R"%9 | | : . | 78$gene | | |