

Summary table of the Rare Disease Genomic Test Logistics for the South West

<ul style="list-style-type: none"> • Core Rare disease tests • Specialised rare disease tests: <ul style="list-style-type: none"> – neurology – cardiac – renal • Whole Genome Sequencing tests 	<p>Bristol Genetics Laboratory Pathology Sciences Southmead Hospital Westbury-on-Trym Bristol BS10 5NB</p>
<ul style="list-style-type: none"> • Specialised rare disease tests: <ul style="list-style-type: none"> – haematology – ophthalmology – gastroenterology – hearing – immunology – inherited cancer – metabolic – mitochondrial – musculoskeletal – respiratory – skin – non-invasive pre-natal diagnosis 	<p>Bristol Genetics Laboratory Pathology Sciences Southmead Hospital Westbury-on-Trym Bristol BS10 5NB</p> <p>DNA WILL BE EXTRACTED FROM SAMPLES AND FORWARDED TO ANOTHER LABORATORY IN THE GLH NETWORK.</p>
<ul style="list-style-type: none"> • <i>HFE</i> related haemochromatosis (R95) • Specialist rare disease tests for endocrine disorders • Exome sequencing service (R14 and R89) 	<p>Exeter Genomics Laboratory RILD level 3 Royal Devon and Exeter NHS Foundation Trust Barrack Road Exeter EX2 5DW</p>