

Exons Not Part of NGS Testing

The indicated exons in the following genes are technically challenging to sequence with NGS and Sanger technologies. Variation cannot be accurately detected in these exons, and therefore they are not analyzed as part of routine NGS testing and backfill sequencing.

Connective Tissue Disorders Panels		
Subpanel: Ehlers-Danlos Syndrome		
Gene	Transcript	Exon(s)
<i>TNXB</i>	NM_019105.6	32-44 (inclusive)

Hereditary Hearing Loss Panels		
Subpanel: Common & Non-Syndromic Hearing Loss		
Gene	Transcript	Exon(s)
<i>ESPN</i>	NM_031475.2	4,7
<i>OTOA</i>	NM_144672.3	22-25, 27
<i>PTPRQ</i>	ENST00000266688.5	4-11 (inclusive)
<i>TPRN</i>	NM_001128228.2	1
<i>TRIOBP</i>	NM_001039141.2	7

Hereditary Spastic Paraplegia Panels		
Subpanel: Autosomal Recessive HSP		
Gene	Transcript	Exon(s)
<i>VPS37A</i>	NM_152415.2	3,4
<i>HSPD1</i>	NM_002156.4	6
<i>KIF1C</i>	NM_006612.5	23