

TABLE E-1 Genetic Findings

Patient	Gene	Mutation
1	<i>EXT2</i>	c.200dupC (p.val68Glyfs*3)
2	<i>EXT1</i>	c.1280T>C (p.Leu427Pro)
3	<i>EXT1</i>	c.649-665del (p.Ala217Tyrfs*2)
4	<i>EXT2</i>	c.626+1G>A
5	<i>EXT1</i>	c.1418-2A>G
6	<i>EXT2</i>	c.1197C>G (p.Tyr399*)
7	<i>EXT2</i>	c.1080-2A>G