

XIV. Appendix G

Denaturing high-performance liquid chromatography conditions

The flow rate of the acetonitrile step gradient, generated by mixing Buffers A and B, was 0.9 ml/min. The percentage of Buffer A is given at time points 0.0, 0.1, 2.1, 2.2, 2.3, 2.4 and 2.5 minutes (Table XIV-1). The percentage of Buffer B can be calculated as follows:

$$B (\%, v/v) = 100\% (v/v) - A (\%, v/v)$$

Table XIV-1 DHPLC conditions for <i>hKIAA1202</i> mutation screening										
Amplicon	T1 (°C)	A (% v/v)	T2 (°C)	A (% v/v)	T3 (°C)	A (% v/v)	T4 (°C)	A (% v/v)		
Exon 1/1	64.9	50.0, 47.0, 37.0, 50.0, 50.0, 50.0, 50.0	66.7	51.0, 48.0, 38.0, 51.0, 51.0, 51.0, 51.0	67.5	52.0, 49.0, 39.0, 52.0, 52.0, 52.0, 52.0	NA	NA		
Exon 1/2	62.3	47.0, 44.0, 34.0, 47.0, 47.0, 47.0, 47.0	64.5	49.0, 46.0, 36.0, 49.0, 49.0, 49.0, 49.0	66.3	51.0, 48.0, 38.0, 51.0, 51.0, 51.0, 51.0	NA	NA		
Exon 1a	54.8	50.0, 47.0, 37.0, 50.0, 50.0, 50.0, 50.0	56.5	52.0, 49.0, 39.0, 52.0, 52.0, 52.0, 52.0	57.9	53.0, 50.0, 40.0, 53.0, 53.0, 53.0, 53.0	NA	NA		
Exon 2	58.5	46.0, 43.0, 33.0, 46.0, 46.0, 46.0, 46.0	60.0	48.0, 45.0, 35.0, 48.0, 48.0, 48.0, 48.0	60.6	49.0, 46.0, 36.0, 49.0, 49.0, 49.0, 49.0	NA	NA		
Exon 2a	54.4	46.0, 43.0, 33.0, 46.0, 46.0, 46.0, 46.0	56.3	48.0, 45.0, 35.0, 48.0, 48.0, 48.0, 48.0	57.3	49.0, 46.0, 36.0, 49.0, 49.0, 49.0, 49.0	58.3	50.0, 47.0, 37.0, 50.0, 50.0, 50.0, 50.0		
Exon 3	60.8	47.0, 44.0, 34.0, 47.0, 47.0, 47.0, 47.0	62.1	48.0, 45.0, 35.0, 48.0, 48.0, 48.0, 48.0	63.4	49.0, 46.0, 36.0, 49.0, 49.0, 49.0, 49.0	NA	NA		

Table XIV-1 DHPLC conditions for <i>hKIAA1202</i> mutation screening									
Amplicon	T1 (°C)	A (% v/v)	T2 (°C)	A (% v/v)	T3 (°C)	A (% v/v)	T4 (°C)	A (% v/v)	
Exon 4/1	60.4	46.0, 43.0, 33.0, 46.0, 46.0, 46.0	61.5	47.0, 44.0, 34.0, 47.0, 47.0, 47.0	62.5	48.0, 45.0, 35.0, 48.0, 48.0, 48.0	NA	NA	
Exon 4/2	62.7	48.0, 45.0, 35.0, 48.0, 48.0, 48.0	64.0	49.0, 46.0, 36.0, 49.0, 49.0, 49.0	NA	NA	NA	NA	
Exon 4/3	61.0	48.0, 45.0, 35.0, 48.0, 48.0, 48.0	62.0	49.0, 46.0, 36.0, 48.0, 48.0, 49.0	64.0	51.0, 48.0, 38.0, 48.0, 48.0, 51.0	NA	NA	
Exon 4/4	60.9	47.0, 44.0, 34.0, 47.0, 47.0, 47.0	61.7	48.0, 45.0, 35.0, 48.0, 48.0, 48.0	NA	NA	NA	NA	
Exon 4/5	60.5	48.0, 45.0, 35.0, 47.0, 47.0, 48.0	62.0	50.0, 47.0, 37.0, 47.0, 47.0, 50.0	63.0	51.0, 48.0, 38.0, 47.0, 47.0, 51.0	NA	NA	
Exon 4/6	60.5	48.0, 45.0, 35.0, 47.0, 47.0, 48.0	61.5	49.0, 46.0, 36.0, 47.0, 47.0, 49.0	NA	NA	NA	NA	
Exon 4/7	62.3	48.0, 45.0, 35.0, 48.0, 48.0, 48.0	63.3	49.0, 46.0, 36.0, 49.0, 49.0, 49.0	64.7	50.0, 47.0, 37.0, 50.0, 50.0, 50.0	NA	NA	
Exon 4/8	59.2	48.0, 45.0, 35.0, 48.0, 48.0, 48.0	60.4	49.0, 46.0, 36.0, 49.0, 49.0, 49.0	61.7	51.0, 48.0, 38.0, 51.0, 51.0, 51.0	NA	NA	
Exon 4/9	60.3	47.0, 44.0, 34.0, 47.0, 47.0, 47.0	61.7	49.0, 46.0, 36.0, 49.0, 49.0, 49.0	62.5	50.0, 47.0, 37.0, 50.0, 50.0, 50.0	NA	NA	
Exon 4/10	57.5	48.0, 45.0, 35.0, 47.0, 47.0, 48.0	60.5	51.0, 48.0, 38.0, 47.0, 47.0, 51.0	61.4	52.0, 49.0, 39.0, 47.0, 47.0, 52.0	NA	NA	
Exon 4/11	57.9	48.0, 45.0, 35.0, 48.0, 48.0, 48.0	59.0	49.0, 46.0, 36.0, 49.0, 49.0, 49.0	59.9	50.0, 47.0, 37.0, 50.0, 50.0, 50.0	NA	NA	
Exon 4/12	57.5	48.0, 45.0, 35.0, 48.0, 48.0, 48.0	58.3	49.0, 46.0, 36.0, 49.0, 49.0, 49.0	59.5	50.0, 47.0, 37.0, 50.0, 50.0, 50.0	NA	NA	

Table XIV-1 DHPLC conditions for <i>hKIAA1202</i> mutation screening									
Amplicon	T1 (°C)	A (% v/v)	T2 (°C)	A (% v/v)	T3 (°C)	A (% v/v)	T4 (°C)	A (% v/v)	
Exon 4/13	58.1	49.0, 46.0, 36.0, 48.0, 48.0, 49.0, 49.0	61.3	52.0, 49.0, 39.0, 48.0, 48.0, 52.0, 52.0	62.4	53.0, 50.0, 40.0, 48.0, 48.0, 53.0, 53.0	NA	NA	
Exon 5	58.0	49.0, 46.0, 36.0, 48.0, 48.0, 49.0, 49.0	60.3	51.0, 48.0, 38.0, 48.0, 48.0, 51.0, 51.0	62.5	58.0, 55.0, 45.0, 53.0, 53.0, 58.0, 58.0	NA	NA	
Exon 6/1a	58.7	50.0, 47.0, 37.0, 50.0, 50.0, 50.0, 50.0	59.5	51.0, 48.0, 38.0, 50.0, 50.0, 51.0, 51.0	61.0	55.0, 52.0, 42.0, 52.0, 52.0, 55.0, 55.0	NA	NA	
Exon 6/1b	59.3	49.0, 46.0, 36.0, 48.0, 48.0, 49.0, 49.0	62.5	54.0, 51.0, 41.0, 52.0, 52.0, 54.0, 54.0	64.5	56.0, 53.0, 43.0, 54.0, 54.0, 56.0, 56.0	NA	NA	
Exon 6/3	57.7	46.0, 43.0, 33.0, 46.0, 46.0, 46.0, 46.0	60.8	47.0, 44.0, 34.0, 47.0, 47.0, 47.0, 47.0	61.3	48.0, 45.0, 35.0, 48.0, 48.0, 48.0, 48.0	NA	NA	
Exon 7	56.2	46.0, 43.0, 33.0, 46.0, 46.0, 46.0, 46.0	59.0	49.0, 46.0, 36.0, 46.0, 46.0, 49.0, 49.0	59.5	49.0, 46.0, 36.0, 46.0, 46.0, 49.0, 49.0	NA	NA	
Exon 8/1a	58.7	52.0, 49.0, 39.0, 52.0, 52.0, 52.0, 52.0	59.3	53.0, 50.0, 40.0, 52.0, 52.0, 53.0, 53.0	64.2	66.0, 63.0, 53.0, 58.0, 58.0, 66.0, 66.0	NA	NA	
Exon 8/1b	58.3	52.0, 49.0, 39.0, 51.0, 51.0, 52.0, 52.0	59.7	53.0, 50.0, 40.0, 51.0, 51.0, 53.0, 53.0	61.9	55.0, 52.0, 42.0, 51.0, 51.0, 55.0, 55.0	63.5	57.0, 54.0, 44.0, 51.0, 51.0, 57.0, 57.0	
Exon 8/2	58.2	49.0, 46.0, 36.0, 49.0, 49.0, 49.0, 49.0	61.5	53.0, 50.0, 40.0, 53.0, 53.0, 53.0, 53.0	62.7	54.0, 51.0, 41.0, 54.0, 54.0, 54.0, 54.0	NA	NA	
Exon 9/1	57.5	47.0, 44.0, 34.0, 47.0, 47.0, 47.0, 47.0	60.5	48.0, 45.0, 35.0, 48.0, 48.0, 48.0, 48.0	63.3	52.0, 49.0, 39.0, 52.0, 52.0, 52.0, 52.0	64.0	53.0, 50.0, 40.0, 53.0, 53.0, 53.0, 53.0	
Exon 9/2	58.0	48.0, 45.0, 35.0, 47.0, 47.0, 48.0, 48.0	60.8	52.0, 49.0, 39.0, 51.0, 51.0, 52.0, 52.0	NA	NA	NA	NA	
Exon 9/3	57.3	47.0, 44.0, 34.0, 47.0, 47.0, 47.0, 47.0	60.6	49.0, 46.0, 36.0, 49.0, 49.0, 49.0, 49.0	61.6	50.0, 47.0, 37.0, 50.0, 50.0, 50.0, 50.0	NA	NA	

Table XIV-1 DHPLC conditions for <i>hKIAA1202</i> mutation screening									
Amplicon	T1 (°C)	A (% v/v)	T2 (°C)	A (% v/v)	T3 (°C)	A (% v/v)	T4 (°C)	A (% v/v)	
Exon 9/4	60.0	46.0, 43.0, 33.0, 45.0, 45.0, 46.0, 46.0	61.5	50.0, 47.0, 37.0, 47.0, 47.0, 50.0, 50.0	62.3	51.0, 48.0, 38.0, 48.0, 48.0, 51.0, 51.0	NA	NA	
Exon 9/5	58.3	47.0, 44.0, 34.0, 47.0, 47.0, 47.0, 47.0	59.9	49.0, 46.0, 36.0, 48.0, 48.0, 49.0, 49.0	60.3	50.0, 47.0, 37.0, 49.0, 49.0, 50.0, 50.0	NA	NA	
Exon 9/6	57.6	52.0, 49.0, 39.0, 52.0, 52.0, 52.0, 52.0	59.6	54.0, 51.0, 41.0, 54.0, 54.0, 54.0, 54.0	61.8	58.0, 55.0, 45.0, 58.0, 58.0, 58.0, 58.0	NA	NA	
Exon 9/7	52.8	46.0, 43.0, 33.0, 46.0, 46.0, 46.0, 46.0	53.4	47.0, 44.0, 34.0, 47.0, 47.0, 47.0, 47.0	58.9	52.0, 49.0, 39.0, 52.0, 52.0, 52.0, 52.0	60.3	53.0, 50.0, 40.0, 52.0, 52.0, 53.0, 53.0	