

**Supplementary Table 1**

Summary of variations identified in 14 individuals

Individual	Known variations								Unknown variations								Total		
	Exon							Sub-total	Exon							Sub-total			
	Single variation				Indel				Splice +/- 2	Single variation				Indel				Splice +/- 2	
	Synonym	Missense	Stop loss	Nonsense	Fs	N Fs	Synonym			Missense	Stop loss	Nonsense	Fs	N Fs					
<b>ALZ 049</b>	7739	6301	9	30	21	19	78	<b>14197</b>	347	567	0	14	60	57	9	<b>1054</b>	<b>15251</b>		
<b>ALZ 426</b>	8030	6534	7	30	20	19	76	<b>14716</b>	333	526	1	12	62	54	6	<b>994</b>	<b>15710</b>		
<b>ROU 632</b>	8040	6540	3	34	19	18	82	<b>14736</b>	323	527	2	20	54	71	19	<b>1016</b>	<b>15752</b>		
<b>EXT 049</b>	8060	6696	5	29	18	22	68	<b>14898</b>	382	602	1	14	71	74	14	<b>1158</b>	<b>16056</b>		
<b>EXT 055</b>	7747	6210	7	32	19	21	68	<b>14104</b>	359	623	0	23	65	59	13	<b>1142</b>	<b>15246</b>		
<b>ALZ 062</b>	7876	6385	7	33	22	18	74	<b>14415</b>	345	594	1	13	71	58	6	<b>1088</b>	<b>15503</b>		
<b>ROU 816</b>	8011	6527	5	39	15	23	81	<b>14701</b>	362	587	1	13	73	57	11	<b>1104</b>	<b>15805</b>		
<b>ALZ 198</b>	7282	5930	5	27	19	22	66	<b>13351</b>	314	575	1	12	56	71	10	<b>1039</b>	<b>14390</b>		
<b>ALZ 056</b>	7860	6592	5	33	22	14	74	<b>14600</b>	280	522	2	15	40	57	18	<b>934</b>	<b>15534</b>		
<b>EXT 094</b>	8300	6837	5	41	19	19	91	<b>15312</b>	338	563	2	10	49	56	10	<b>1028</b>	<b>16340</b>		
<b>EXT 077</b>	8050	6641	7	39	23	17	74	<b>14851</b>	309	478	2	9	53	51	9	<b>911</b>	<b>15762</b>		
<b>EXT 050</b>	8156	6683	7	27	21	20	75	<b>14989</b>	274	459	0	10	53	58	15	<b>869</b>	<b>15858</b>		
<b>EXT 220</b>	10070	8558	26	36	21	19	94	<b>18824</b>	362	585	1	2	152	115	14	<b>1231</b>	<b>20055</b>		
<b>EXT 181</b>	9981	8487	27	30	19	16	84	<b>18644</b>	373	681	3	4	167	107	22	<b>1357</b>	<b>20001</b>		

**Legend:** Fs: Frameshift N Fs: Not Frameshift