

Table I – Coding Region sequences between positions 3230-4331 (Andrews et al. 1999) of the Azorean families analysed. Data from Santos et al (2005), for the D-loop region, are also presented.

Family	Samples	Coding Region tRNA <sup>Leu</sup> , ND1 and tRNA <sup>Ile</sup> (3230-4331)	D-loop (16024-16569 and 1-400) (Santos et al 2005)	Haplogroup
AC_I	III1, 2, 3, 4, 5	-	16519, 152, 263, 311.1	H or HV, preHV
AC_II	II3, III1	-	16519, 263, 311.1	H or HV, preHV
AV	III9, IV1, 2, 5, 6, 7, 8, 9	-	16291, 16298, 72, 195, 263, <i>Het Poly-C 303-309 (303.1, 303.2, 303.3)</i> , 311.1	V or pre*V
	IV3, 4	<b>Het 3260 A/G</b>	16291, 16298, 72, 195, 263, <i>Het Poly-C 303-309 (303.1, 303.2, 303.3)</i> , 311.1	
	III3	-	Not Analysed	
	III1	-	-	
BF	III,2,3,4,5	4216, 4232	16069, 16126, 16278, 16366, 16519, 73, 185, 188, 228, 263, 295, <i>Het Poly-C 303-309 (303.1, 303.2, 303.3)</i> , 311.1 [462, 489]	J
BM	III1, 4, IV1, 2	3348, 3969	16108, 16111, 16172, 16189, <i>Het Poly-C 16184-16193 (16190.0, 16190.1, 16190.2)</i> , 16219, 16278, 16519, 73, 152, 251, 263, 311.1	U6a1
CAM_I	II3, IV2, 3, 4, 5	3338	16224, 16270, 16T, 73, 150, 199, 263, 279, 311.1, [518T]	U5
	IV1, 6		16224, 16270, 16T, 73, 150, 199, 263, 279, <i>Het Poly-C 303-309 (303.0, 303.1)</i> , 311.1, [518T]	
CAM_II	III, 2	-	16114A, 16192, 16256, 16270, 16294, 16526, 73, 195, 263, <i>Het Poly-C 303-309 (303.1, 303.2)</i> , 311.1	U5a1
CAM_III	III1	-	16239G, 16256, 16519, 152, 263, <i>Het Poly-C 303-309 (303.0, 303.1, 303.2, 303.3)</i> , 311.1	H or HV, preHV
	III2		16239G, 16256, 16519, 152, 263, <i>Het Poly-C 303-309 (303.1, 303.2, 303.3)</i> , 311.1	
CAR_I	III1, V2, 3, 6, 7, VII	-	16274, 16294G, 16519, 152, 263, 311.1	H or HV, preHV
	V1		<b>Het 16189 T/C</b> , <i>Het Poly-C 16184-16193 (16190.0, 16190.1)</i> , 16274, 16294G, 16519, 152, 263, 311.1	
	IV1	Not Analysed	16274, 16294G, 16519, 152, 263, 311.1	
	IV2	-	16086, 16256, 16270, 16342, 16399 (Only HVRI)	U5a1a

<b>CAR_II</b>	II3	-	16519, 195, 263, <i>Het Poly-C 303-309 (303.1, 303.2, 303.3)</i> , 311.1	H or HV, preHV
	III1		16519, 195, 263, <i>Het Poly-C 303-309 (303.1, 303.2)</i> , 311.1	
	II4		16519, <b>Het 64 T/C</b> , 195, 263, <i>Het Poly-C 303-309 (303.1, 303.2)</i> , 311.1	
	I2	Not Analysed	16519, 195, 263, <i>Het Poly-C 303-309 (303.1, 303.2, 303.3)</i> , 311.1	
	I1		152, 263, 311.1 (Only HVR II)	
<b>CAR_III</b>	III, 3, 4	-	16086, 16104, 16183C, 16189, <i>Het Poly-C 16184-16193 (16190.1, 16190.2)</i> , 16223, 16271, 16278, 16519, 73, 153, 195, 225, 226, 263, <i>Het Poly-C 303-309 (303.1, 303.2, 303.3)</i> , 311.1	X
	II2		16086, 16104, 16183C, 16189, <i>Het Poly-C 16184-16193 (16190.0, 16190.1, 16190.2)</i> , 16223, 16271, 16278, 16519, 73, 153, 195, 225, 226, 263, <i>Het Poly-C 303-309 (303.1, 303.2)</i> , 311.1	
<b>CAR_IV</b>	III, 2	-	16274, 16294G, 16519, 152, 263, 311.1	H or HV, preHV
<b>CAR_V</b>	III1, 2, 3, 4, 5, 6, 7, 8	4216, 4232	16069, 16126, 16278, 16366, 16519, 73, 185, 188, 228, 263, 295, <i>Het Poly-C 303-309 (303.1, 303.2, 303.3)</i> , 311.1 [465, 492]	J
<b>CAR_VI</b>	III, 2	-	16519, 263, 311.1	H or HV, preHV
<b>CG_I</b>	II3, III3, 4, 5, IV1	3421, 3594, 3666, 3777, 4104	16129, 16187, 16189, 16223, 16265C, 16278, 16286A, 16292, 16294, 16311, 16360, 16519, 16527, 73, 152, 182, 186A, 189C, 195, 198, 247, 263, 297, 311.1, 316	L1c2
<b>CG_II</b>	III, 4	-	16298, 72, 195, 263, <i>Het Poly-C 303-309 (303.1, 303.2, 303.3)</i> , 311.1	V or pre*V
	II2	<b>Het 3396 T/C</b>	16298, 72, 195, 263, <i>Het Poly-C 303-309 (303.1, 303.2, 303.3)</i> , 311.1	
	III, 5	-	16298, 72, 195, 263, <i>Het Poly-C 303-309 (303.1, 303.2)</i> , 311.1	
	I2	-	Not Analysed	Not Analysed
I1	3421, 3594, 3666, 3777, 4104	Not Analysed		
<b>CG_III</b>	III,4	-	16519, 199, 263, <i>Het Poly-C 303-309 (303.1, 303.2)</i> , 311.1	H or HV, preHV
	II2,3		16519, 199, 263, <i>Het Poly-C 303-309 (303.1, 303.2, 303.3)</i> , 311.1	
<b>CM</b>	IV1, 2	3992, 4024	16265C, 263, <i>Het Poly-C 303-309 (303.1, 303.2, 303.3)</i> , 311.1	H or HV, preHV
	IV3, 5		16265C, 263, <i>Het Poly-C 303-309 (303.0, 303.1, 303.2)</i> , 311.1	
	IV4		16265C, 263, <i>Het Poly-C 303-309 (303.1, 303.2)</i> , 311.1	
<b>CV</b>	IV1, 2, 3, 4	3420, 3450	16124, 16171, 16183C, 16189, <i>Het Poly-C 16184-16193 (16190.0, 16190.1, 16190.2)</i> , 16214, 16223, 16278, 16362, 73, 263, <i>Het Poly-C 303-309 (303.1, 303.2, 303.3)</i> , 311.1	L3b
<b>ER</b>	III, 2, 3	3394, 4216	16069, 16126, 16221, 73, 185, 228, 263, 295, 303.1, 311.1 [464, 484, 491]	J
<b>HR_I</b>	III1, 2, 7, IV1	-	16192, 16260, 16519, 263, 311.1	H or HV, preHV
	IV2		<b>Het 16189 T/C</b> , <i>Het Poly-C 16184-16193 (16190.0, 16190.1)</i> , 16192, 16260, 16519, 263, 311.1	
	II2, 3, III5	Not Analysed	<b>Het 16189 T/C</b> , <i>Het Poly-C 16184-16193 (16190.0, 16190.1)</i> , 16192, 16260, 16519, 263, 311.1	

<b>HR_II</b>	III1	3796	16519, 93, 263, <i>Het Poly-C 303-309 (303.1, 303.2, 303.3)</i> , 311.1	H or HV, preHV
	III2, IV1, 2, 3		16519, 93, 263, <i>Het Poly-C 303-309 (303.1, 303.2)</i> , 311.1	
<b>JA_I</b>	III9, IV1, 2, 3, 4, 5, 6, 7, 8, 9, 10	4216, 4232	16069, 16126, 16278, 16366, 16519, 73, 185, 188, 228, 263, 295, <i>Het Poly-C 303-309 (303.1, 303.2)</i> , 311.1 [462, 489]	J
	III10		16069, 16126, 16278, 16366, 16519, 73, 185, 188, 228, <b>Het 238 A/G</b> , 263, 295, <i>Het Poly-C 303-309 (303.1, 303.2)</i> , 311.1 [462, 489]	
<b>JA_II</b>	II3, III2	-	16293, 16311, 16519, 146, 195, 263, <i>Het Poly-C 303-309 (303.1, 303.2, 303.3)</i> , 311.1	H or HV, preHV
	III1		16293, 16311, 16519, 146, 195, 263, <i>Het Poly-C 303-309 (303.1, 303.2)</i> , 311.1	
<b>JA_III</b>	III, 2	-	16183C, 16189, <i>Het Poly-C 16184-16193 (16190.0, 16190.1, 16190.2)</i> , 16519, 187T, 263, 311.1	H or HV, preHV
<b>JL_I</b>	III, 2	-	16519, 263, <i>Het Poly-C 303-309 (303.1, 303.2)</i> , 311.1 [479]	H or HV, preHV
<b>JL_II</b>	III, 2	-	16519, 263, <i>Het Poly-C 303-309 (303.1, 303.2)</i> , 311.1	H or HV, preHV
<b>JS</b>	II5, III1, 2	3505, <b>Het 3602 A/G</b>	16223, 16292, 16320, 16519, 73, 189, 195, 204, 207, 263, <i>Het Poly-C 303-309 (303.0, 303.1)</i> , 311.1	W
	III3	3505	16223, 16292, 16320, 16519, 73, 189, 195, 204, 207, 263, <i>Het Poly-C 303-309 (303.0, 303.1)</i> , 311.1	
	I2, II2, 3	3505, <b>Het 3602 A/G</b>	Not Analysed	
<b>MA_I</b>	III1, IV1, 2	4117	16519, 263, 311.1	H or HV, preHV
<b>MA_II</b>	III, 2	-	16252, 16270, 16342, 16399, 73, 200, 263, 311.1	U5
<b>MA_III</b>	III, 2	-	16519, 152, 263, 311.1	H or HV, preHV
<b>MBP</b>	III3, IV1, 5	-	16129, 16183C, 16189, <i>Het Poly-C 16184-16193 (16190.0, 16190.1, 16190.2)</i> , 16223, 16249, 16311, 16399, 16454, 73, 195, 263, <i>Het Poly-C 303-309 (303.1, 303.2, 303.3)</i>	M1
	IV2, 3		16129, 16183C, 16189, <i>Het Poly-C 16184-16193 (16190.1, 16190.2)</i> , 16223, 16249, 16311, 16399, 16454, 73, 195, 263, <i>Het Poly-C 303-309 (303.1, 303.2)</i>	
	IV4		16129, 16183C, 16189, <i>Het Poly-C 16184-16193 (16190.1, 16190.2)</i> , 16223, 16249, 16311, 16399, 16454, 73, 195, 263, <i>Het Poly-C 303-309 (303.0, 303.1, 303.2, 303.3)</i>	
<b>ML</b>	III1	Not Analysed	16129, 16183C, 16189, <i>Het Poly-C 16184-16193 (16190.0, 16190.1, 16190.2)</i> , 16249, 16311, 16519, 73, 263, <i>Het Poly-C 303-309 (303.1, 303.2)</i> , 311.1 [489]	M1
	III3		16129, 16183C, 16189, <i>Het Poly-C 16184-16193 (16190.0, 16190.1, 16190.2, 16190.3)</i> , 16249, 16311, 16519, 73, 263, 311.1 [489]	
	III2		<b>Het 16086 T/C</b> , 16129, <b>Het 16182 C/A or del A</b> , 16183C, 16189, <i>Het Poly-C 16184-16193 (16190.-2, 16190.-1, 16190.0, 16190.1, 16190.2, 16190.3, 16190.4, 16190.5)*</i> , 16249, 16311, 16519, 73, 263, <i>Het Poly-C 303-309 (303.0, 303.1, 303.2, 303.3, 303.4)</i> , 311.1, [489]	
	II4		<b>Het 16086 T/C</b> , 16129, <b>Het 16182 C/A or del A</b> , 16183C, 16189, <i>Het Poly-C 16184-16193 (16190.-2, 16190.-1, 16190.0, 16190.1, 16190.2, 16190.3, 16190.4, 16190.5)*</i> , 16249, 16311, 16519, 73, 263, 303.3, 311.1, <i>Het Poly-C 303-309 (303.0, 303.1, 303.2, 303.3, 303.4)</i> [489]	
	II2		16129, 16183C, 16189, <i>Het Poly-C 16184-16193 (16190.-1, 16190.0, 16190.1, 16190.2, 16190.3)</i> , 16249, 16311, 16519, 73, 263, 303.2, 311.1, <i>Het Poly-C 303-309 (303.1, 303.2, 303.3)</i> [489]	

MM_I	III, III, 2	3434, 4216	16069, 16126, 16145, 16231, 16261, 73, 150, 152, 195, 215, 263, 295, 309.1T, 311.1, 319	J1a
	II2		16069, 16126, 16145, 16231, 16261, <b>Het Poly G 66-71 delG</b> , 73, 150, 152, 195, 215, 263, 295, 309.1T, 311.1, 319	
	I2	Not Analysed	16069, 16126, 16145, 16231, 16261, <b>Het Poly G 66-71 delG</b> , 73, 150, 152, 195, 215, 263, 295, 309.1T, 311.1, 319	
MM_II	III, 2	3918	16293, 16311, 195, 263, <i>Het Poly-C 303-309 (303.1, 303.2)</i> , 311.1	H or HV, preHV
MN	III1, 2, 3, 4	-	16189, <i>Het Poly-C 16184-16193 (16190.0, 16190.1, 16190.2, 16190.3)</i> , 16298, 72, 263, <i>Het Poly-C 303-309 (303.1, 303.2, 303.3)</i> , 311.1	V or pre*V
NP_I	III1, 2, 3, 4, 5	3348	16172, 16183C, 16189, <i>Het Poly-C 16184-16193 (16190.1, 16190.2)</i> , 16219, 16278, 73, 263, 311.1	U6a
NP_II	III8, IV1, 2, 3	4216	16069, 16126, 16150, 16519, 73, 185, 188, 228, 263, 295, 311.1 [463, 490]	J
	III1		16069, 16126, <b>16150 Het T/C</b> , 16519, 73, 185, 188, 228, 263, 295, 311.1 [463, 490]	
	II2	Not Analysed	16069, 16126, <b>16150 Het T/C</b> , 16519, 73, 185, 188, 228, 263, 295, 311.1 [463, 490]	
	III6		16069, 16126, 16150, 16519, 73, 185, 188, 228, 263, 295, 311.1 [463, 490]	
NP_III	IV4, V1	3480	16224, 16311, 16519, 73, 195, 263, 311.1 [498]	K
	IV1		16224, <b>16309 Het A/G</b> , 16311, 16519, 73, 195, 263, 315 (1C) [498]	
	III2	Not Analysed	16224, 16311, 16519, 73, 195, 263, 315 (1C) [498]	
	III1		16519 (Only HVRI)	H or HV, preHV
	IV7, V2, VII	3480	16224, 16311, 16519, 73, <b>150 Het C/T</b> , 195, 263, 311.1 [498]	K
	V3	Not Analysed	16224, 16311, 16519, 73, <b>150 Het C/T</b> , 195, 263, 311.1 [498]	
	IV6		16224, 16311, 16519, 73, <b>150 Het C/T</b> , 195, 263, 311.1 [498]	
III5	16224, 16311, 16519, 73, <b>150 Het C/T</b> , 195, 263, 311.1 [498]			
NP_IV	III, 2, 3	-	16140, 16519, 263, <i>Het Poly-C 303-309 (303.1, 303.2, 303.3)</i> , 311.1	H or HV, preHV
NP_V	III, 2, 3	-	16304, 185, 263, 311.1 [456]	H or HV, preHV
PA_I	IV1, 2, 3	-	16129, 16183C, 16189, <i>Het Poly-C 16184-16193 (16190.0, 16190.1, 16190.2)</i> , 16249, 16311, 16355, 16519, 73, 263, <i>Het Poly-C 303-309 (303.1, 303.2)</i> , 311.1	U1
	IV4		16129, 16183C, 16189, <i>Het Poly-C 16184-16193 (16190.0, 16190.1, 16190.2)</i> , 16249, <b>16264 Het T/C</b> , 16311, 16355, 16519, 73, 263, <i>Het Poly-C 303-309 (303.1, 303.2)</i> , 311.1	
	III5	Not Analysed	16129, 16183C, 16189, <i>Het Poly-C 16184-16193 (16190.0, 16190.1, 16190.2)</i> , 16249, 16311, 16355, 16519, 73, 263, <i>Het Poly-C 303-309 (303.1, 303.2)</i> , 311.1	
	III6		16069, 16126, 16278, 16366, 16519 (Only HVRI)	J

<b>PA_II</b>	II3	-	16519, 263, <i>Het Poly-C 303-309 (303.1, 303.2)</i> , 311.1	H or HV, preHV
	III,2		16519, <b>152 Het T/C</b> , 263, <i>Het Poly-C 303-309 (303.1, 303.2, 303.3)</i> , 311.1	
	I2	Not Analysed	16519, <b>152 Het T/C</b> , 263, <i>Het Poly-C 303-309 (303.1, 303.2)</i> , 311.1	
<b>PP</b>	III1, 2, 3, 4, 5, 6	3594, 4104	16223, 16278, 16294, 16309, 16390, 73, 143, 152, 195, 263, <i>Het Poly-C 303-309 (303.1, 303.2)</i> , 311.1	L2a
<b>RUB</b>	III,2,3,4,5	-	16519, 263, 311.1	H or HV, preHV
<b>SAO</b>	III1, 2, 3, 4	Not analysed	16201, 16278, 16519, <b>Het 215 G/A</b> , 263, 311.1	H or HV, preHV
	III		16201, 16278, 16519, <b>Het 215 G/A</b> , 263, 311.1	
	I2		16201, 16278, 16519, <b>Het 215 G/A</b> , 263, 311.1	
<b>TM</b>	II7, 10, 11, III1, 2, 3, 4, 5, 6, 7	3499	16239, 16519, 263, 311.1	H or HV, preHV
<b>ZH</b>	III5, 6, 7, IV1, 2, 3, 4, 5, 6, 7, 8	-	16519, 152, 263, 311.1	H or HV, preHV

Substitutions are transitions unless the base change is explicitly indicated.

\* Variants detected by sequencing of multiple clones.