**Supplementary table 1**

| **Age** | **Sex** | **Height (m)** | **Mass (kg)** | **BMI** | **Heteroplasmy (%)** | **Respiratory capacity (FVC)** | **Cognitive score (ACE-R)** | **Principle clinical features** |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **m.3243A>G Genotype** (mean±SD) | | | | | | | | |
| **58** | **F** | **173** | **52.1** | **17.4** | **59** | **2.55** | **98** | **Hearing loss, ataxia, constipation, underweight, myopathy, myalgia, exercise intolerance** |
| **39** | **M** | **172** | **74.6** | **25.2** | **80** | **4.95** | **93** | **Hearing loss, diabetes, migraine, fatigue, hypothyroidism, ataxia, constipation, exercise intolerance** |
| **37** | **F** | **164** | **50.7** | **18.9** | **48** | **2.8** | **91** | **Hearing loss, diabetes, exercise intolerance, ataxia, dysarthria, asthma** |
| **42** | **M** | **176** | **83.7** | **27.0** | **82** | **3.96** | **90** | **Hearing loss, diabetes, migraine, ataxia, depression, exercise intolerance** |
| **47** | **M** | **182** | **64.0** | **19.3** | **63** | **3.11** | **94** | **Hearing loss, diabetes, fatigue, ataxia, myalgia, depression, myopathy, neuropathy, ptosis, PEO** |
| **38** | **F** | **164** | **54.2** | **20.1** | **53** | **2.6** | **97** | **Hearing loss, migraine, constipation, myopathy, fatigue, exercise intolerance, asthma** |
| **22** | **M** | **183** | **59.2** | **17.7** | **89** | **4.23** | **94** | **Hearing loss, migraine, epilepsy, ataxia, constipation, underweight, exercise intolerance, fatigue** |
| **36** | **M** | **179** | **72.4** | **22.6** | **80** | **4.8** | **95** | **Migraine, exercise intolerance** |
| **50** | **M** | **162** | **64.6** | **24.6** | **87** | **3.26** | **66** | **Hearing Loss, exercise intolerance, ataxia, myopathy, fatigue, depression, retinopathy, epilepsy, encephalopathy, cognitive decline, stroke-like episodes** |
| **55** | **F** | **153** | **45.8** | **19.5** | **68** | **2.52** | **97** | **Hearing loss, diabetes, myopathy, exercise intolerance, ataxia, constipation, depression, retinopathy, PEO, ptosis, short stature, mild dysphagia, hypertension** |
| 42 | F | 154 | 49.0 | 20.7 | 43 | 2.59 | 90 | Hearing loss, diabetes, ataxia, constipation, depression, exercise intolerance, short stature |
| 50 | F | 169 | 63.4 | 22.2 | 34 | 4.01 | 96 | Migraine, fatigue, hypothyroidism, myalgia, constipation, hypertension, dyslipidaemia, coeliac disease |
| 58 | M | 186 | 89.8 | 25.9 | 66 | 2.6 | 87 | Hearing loss, diabetes, ataxia, retinopathy, constipation, hypertension, dysarthria, myopathy, neuropathy, exercise intolerance |
| 53 | F | 164 | 59.1 | 22.0 | 22 | 3.6 | 92 | Hearing loss, ataxia, retinopathy, constipation, myopathy, exercise intolerance |
| 25 | M | 174 | 49.8 | 16.5 | 90 | 3.39 | 89 | Hearing loss, migraine, ataxia, retinopathy, constipation, depression, dysarthria, myopathy, exercise intolerance, short stature, asthma |
| 18 | F | 154 | 39.5 | 16.7 | 59 | 2.62 | 86 | Hearing loss, retinopathy, underweight, myopathy, exercise intolerance, short stature |
| 24 | F | 155 | 64.0 | 26.7 | 72 | 3.31 | 89 | Hearing loss, migraine, ataxia, retinopathy, constipation, depression, dysarthria, myopathy, exercise intolerance, short stature |
| 55 | M | 179 | 104.5 | 32.6 | 76 | 2.52 | 97 | Hearing loss, diabetes, depression |
|  |  |  |  |  |  |  |  |  |
| 42±13 | 9F, 9M | 1.69 ± .11 | 63.4 ± 16.6**\*** | 22.0 ± 4.3**┼** | 65 ± 19 | 3.30±0.8 | 91±7 |  |
| **Control group** (mean±SD) | | | | | | | | |
| 37 ± 12 | 5F, 7M | 1.72 ± .10 | 80.2 ± 14.9 | 27.1 ±4.8 | - |  |  |  |

\**p=.008,┼**p=.005*