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| **Table A.1. Diagnostic tests collected**  |
| **Cardiac function** |
|   | Electrocardiogram |
|   | Tissue Doppler  |
|   | HOLTER 24h |
|   | Exercise stress test  |
|   | Cardiac Magnetic Resonance  |
| **Renal function** |
|   | Renal echography  |
|   | Renal biopsy  |
| **Neurological function** |
|   | Magnetic resonance imaging  |
|   | Neurophysiological study  |
|   | Transcranial doppler  |
| **Ophthalmological tests** |
|   | Slit-lamp evaluation  |
|   | Ocular funduscopy  |
| **ORL tests** |
|   | Audiometry |
|   | Evoked potentials  |
| **Paediatric tests** |
|   | Evaluation of delayed growth and sexual maturation |
|   | Hands radiography |
| **Other tests** |
|   | Respiratory tests  |
|   | Abdominal echography  |
|   | Osteoarticular radiological study  |
|   | Nailfold capillaroscopy  |

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| **Table A.2. Diagnostic tests collected (results)** |
|   |   |   | Date |
|   |   |   | Before diagnosis | At diagnosis (±6months) | After diagnosis | No date |
|   |   |   | N=33 | N=33 | N=33 | N=33 |
| **Cardiac function** |   |   |   |   |   |
| Electrocardiogram | No | n (%) | 24 (72.7) | 32 (97.0) | 17 (51.5) | 31 (93.9) |
|   | Yes | n (%) | 9 (27.3) | 1 (3.0) | 16 (48.5) | 2 (6.1) |
|   | Altered | n (%) | 1 (12.5) | 0 (0.0) | 3 (20.0) | - |
|   | Normal | n (%) | 7 (87.5) | 1 (100) | 12 (80.0) | - |
|   | Missing | N | 1 | 0 | 1 | 2 |
| Tissue Doppler  | No | n (%) | 29 (87.9) | 29 (87.9) | 16 (48.5) | - |
|   | Yes | n (%) | 4 (12.1) | 4 (12.1) | 17 (51.5) | - |
|   | Altered | n (%) | 1 (25.0) | 0 (0.0) | 5 (38.5) | - |
|   | Normal | n (%) | 3 (75.0) | 3 (100) | 8 (61.5) | - |
|   | Missing | N | 0 | 1 | 4 | - |
| HOLTER 24h | No | n (%) | 31 (93.9) | 0 (0.0) | 31 (93.9) | - |
|   | Yes | n (%) | 2 (6.1) | 0 (0.0) | 2 (6.1) | - |
|   | Altered | n (%) | 0 (0.0) | 0 (0.0) | 2 (100) | - |
|   | Normal | n (%) | 2 (100) | 0 (0.0) | 0 (0.0) | - |
|   | Missing | n | 0 | 0 | 0 | - |
| Exercise stress test  | No | n (%) | 33 (100) | 33 (100) | 33 (100) | - |
|   | Yes | n (%) | 0 (0.0) | 0 (0.0) | 0 (0.0) | - |
| Cardiac Magnetic Resonance  | No | n (%) | 30 (90.9) | 0 (0.0) | 30 (90.9) | - |
| Yes | n (%) | 3 (9.1) | 0 (0.0) | 3 (9.1) | - |
|   | Altered | n (%) | 0 (0.0) | - | 0 (0.0) | - |
|   | Normal | n (%) | 3 (100) | - | 1 (100) | - |
|   | Missing | n | 0 | - | 2 | - |
| **Renal function** |   |   |   |   |   |
| Renal echography  | No | n (%) | 31 (93.9) | 33 (100) | 23 (69.7) | - |
|   | Yes | n (%) | 2 (6.1) | 0 (0.0) | 10 (30.3) | - |
|   | Altered | n (%) | 0 (0.0) | - | 1 (12.5) | 0 (0.0) |
|   | Normal | n (%) | 1 (100) | - | 7 (87.5) | 0 (0.0) |
|   | Missing | n | 1 | - | 2 | 0 |
| Renal biopsy  | No | n (%) | 33 (100) | 33 (100) | 33 (100) | - |
|   | Yes | n (%) | 0 (0.0) | 0 (0.0) | 0 (0.0) | - |
| **Neurological function** |   |   |   |   |   |
| Magnetic resonance imaging  | No | n (%) | 27 (81.8) | 33 (100) | 26 (78.8) | - |
| Yes | n (%) | 6 (18.2) | 0 (0.0) | 7 (21.2) | - |
|   | Altered | n (%) | 1 (16.7) | - | 1 (14.3) | - |
|   | Normal | n (%) | 5 (83.3) | - | 6 (85.7) | - |
|   | Missing | n | 0 | - | 0 | - |
| Neurophysiological study  | No | n (%) | 32 (97.0) | 33 (100) | 23 (69.7) | - |
| Yes | n (%) | 1 (3.0) | 0 (0.0) | 10 (30.3) | - |
|   | Altered | n (%) | 0 (0.0) | 0 (0.0) | 3 (42.9) | - |
|   | Normal | n (%) | 1 (100) | 0 (0.0) | 4 (57.1) | - |
|   | Missing | n | 0 | 0 | 3 | - |
| Transcranial doppler  | No | n (%) | 33 (100) | 33 (100) | 31 (93.9) | - |
|   | Yes | n (%) | 0 (0.0) | 0 (0.0) | 2 (6.1) | - |
|   | Altered | n (%) | - | - | 2 (100) | - |
|   | Normal | n (%) | - | - | 0 (0.0) | - |
|   | Missing | n | - | - | 0 | - |
| **Ophthalmological tests** |   |   |   |   |   |
| Slit-lamp evaluation  | No | n (%) | 25 (75.8) | 33 (100) | 20 (60.6) | 32 (97.0) |
|   | Yes | n (%) | 8 (24.2) | 0 (0.0) | 13 (39.4) | 1 (3.0) |
|   | Altered | n (%) | 1 (14.3) | - | 1 (9.1) | 1 (100) |
|   | Normal | n (%) | 6 (85.7) | - | 10 (90.9) | 0 (0.0) |
|   | Missing | n | 1 | - | 2 | 0 |
| Ocular funduscopy  | No | n (%) | 28 (84.8) | 33 (100) | 18 (54.5) | 32 (97.0) |
|   | Yes | n (%) | 5 (15.2) | 0 (0.0) | 15 (45.5) | 1 (3.0) |
|   | Altered | n (%) | 0 (0.0) | - | 1 (8.3) | 0 (0.0) |
|   | Normal | n (%) | 4 (100) | - | 11 (91.7) | 1 (100) |
|   | Missing | n | 1 | - | 3 | 0 |
| **ORL tests** |   |   |   |   |   |
| Audiometry | No | n (%) | 29 (87.9) | 33 (100) | 20 (60.6) | 32 (97.0) |
|   | Yes | n (%) | 4 (12.1) | 0 (0.0) | 13 (39.4) | 1 (3.0) |
|   | Altered | n (%) | 0 (0.0) | - | 0 (0.0) | - |
|   | Normal | n (%) | 2 (100) | - | 9 (100) | - |
|   | Missing | n | 2 | - | 4 | 1 |
| Evoked potentials  | No | n (%) | 33 (100) | 33 (100) | 33 (100) | 32 (97.0) |
|   | Yes | n (%) | 0 (0.0) | 0 (0.0) | 0 (0.0) | 1 (3.0) |
|   | Altered | n (%) | - | - | - | - |
|   | Normal | n (%) | - | - | - | - |
|   | Missing | n | - | - | - | 1 |
| **Paediatric tests** |   |   |   |   |   |
| Evaluation of delayed growth and sexual maturation | No | n (%) | 33 (100) | 33 (100) | 33 (100) | - |
| Yes | n (%) | 0 (0.0) | 0 (0.0) | 0 (0.0) | - |
| Hands radiography | No | n (%) | 33 (100) | 33 (100) | 31 (93.9) | - |
|   | Yes | n (%) | 0 (0.0) | 0 (0.0) | 2 (6.1) | - |
|   | Altered | n (%) | - | - | 0 (0.0) | - |
|   | Normal | n (%) | - | - | 1 (100) | - |
|   | Missing | n | - | - | 1 | - |
| **Other tests** |   |   |   |   |   |
| Respiratory tests  | No | n (%) | 33 (100) | 33 (100) | 32 (97.0) | - |
|   | Yes | n (%) | 0 (0.0) | 0 (0.0) | 1 (3.0) | - |
|   | Altered | n (%) | - | - | 0 (0.0) | - |
|   | Normal | n (%) | - | - | 1 (100) | - |
|   | Missing | n | - | - | 0 | - |
| Abdominal echography  | No | n (%) | 31 (93.9) | 32 (97.0) | 26 (78.8) | - |
|   | Yes | n (%) | 2 (6.1) | 1 (3.0) | 7 (21.2) | - |
|   | Altered | n (%) | 0 (0.0) | 0 (0.0) | 1 (20.0) | - |
|   | Normal | n (%) | 2 (100) | 1 (100) | 4 (80.0) | - |
|   | Missing | n | 0 | 0 | 2 | - |
| Osteoarticular radiological study  | No | n (%) | 31 (93.9) | 33 (100) | 29 (87.9) | - |
| Yes | n (%) | 2 (6.1) | 0 (0.0) | 4 (12.1) | - |
|   | Altered | n (%) | 1 (100) | - | 1 (25.0) | - |
|   | Normal | n (%) | 0 (0.0) | - | 3 (75.0) | - |
|   | Missing | n | 1 | - | 0 | - |
| Nailfold capillaroscopy  | No | n (%) | 33 (100) | 33 (100) | 33 (100) | - |
|   | Yes | n (%) | 0 (0.0) | 0 (0.0) | 0 (0.0) | - |