|  |  |
| --- | --- |
| **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 |

|  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **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) | | - |