**Supplementary Material**

|  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Table 1S. Comparison of demographic parameter between atypical (GTF2IRD2 deleted) and typical (GTF2IRD2 retained) WS group** | | | | | | | | | | |
|
| **Variable** | **Typical group** | | |  | **Atypical group** | | | ***Mann Whitney U*** | | ***Cohen’s***  ***d*** |
| **M** | **SD** | **Mean rank** |  | **M** | **SD** | **Mean rank** |
|  | ***U*** | ***p*** |
| Chronological Age | 11.50 | 3.59 | 6.44 |  | 12.00 | 4.97 | 6.63 | 15.50 | .93 | 0.05 |
|  |  |  |  |  |  |  |
| Mental age | 5.63 | 1.41 | 6.38 |  | 6.75 | 3.59 | 6.75 | 15.00 | .93 | 0.10 |
|  |
| Education (years) | 5.38 | 3.25 | 6.31 |  | 6.00 | 3.56 | 6.88 | 14.50 | .81 | 0.15 |
|  |
| FSIQ | 48.75 | 4.92 | 6.06 |  | 55.25 | 13.28 | 7.38 | 12.50 | .57 | 0.35 |
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| **Table 2S. Comparison of WISC-IV/WAIS-IV index scores and subtest between atypical and typical WS group** | | | | | | | | | |
| **Variable** | **Typical group** | | | **Atypical group** | | | **Mann Whitney U** | | ***Cohen’s***  ***d*** |
| **M** | **SD** | **Mean rank** | **M** | **SD** | **Mean rank** | **U** | ***p*** |
| **Comprehension verbal index** | 55.75 | 7.21 | 5.44 | 65.50 | 11.85 | 8.63 | 7.50 | .15 | 0.92 |
| Similarities | 3.63 | 1.92 | 5.69 | 5.25 | 2.22 | 8.13 | 9.50 | .28 | 0.67 |
| Vocabulary | 2.38 | 1.06 | 5.50 | 4.00 | 2.45 | 8.50 | 8.00 | .21 | 0.85 |
| Comprehension | 1.63 | 1.77 | 5.88 | 1.75 | 0.96 | 7.75 | 11.00 | .46 | 0.51 |
| Information | 2.75 | 1.49 | 6.31 | 3.25 | 2.63 | 6.88 | 14.50 | .81 | 0.15 |
| Tracks | 3.75 | 1.49 | 6.56 | 3.00 | 1.73 | 4.50 | 7.50 | .38 | 0.92 |
| **Perceptual reasoning index** | 54.25 | 7.85 | 6.13 | 56.25 | 9.91 | 7.25 | 13.00 | .68 | 0.30 |
| Block design | 2.00 | 1.41 | 6.94 | 1.50 | 1.00 | 5.63 | 12.50 | .57 | 0.35 |
| Picture concepts | 1.75 | 1.39 | 6.63 | 2.00 | 2.00 | 6.25 | 15.00 | .93 | 0.10 |
| Matrix reasoning | 3.88 | 2.23 | 6.56 | 4.00 | 2.16 | 6.38 | 15.50 | .93 | 0.05 |
| Picture completion | 2.13 | 1.46 | 5.88 | 3.00 | 1.83 | 7.75 | 11.00 | .46 | 0.51 |
| **Working memory index** | 63.13 | 7.34 | 7.06 | 60.25 | 13.72 | 5.38 | 11.50 | .46 | 0.45 |
| Digit span | 2.63 | 1.92 | 6.31 | 2.75 | 1.71 | 6.88 | 14.50 | .81 | 0.15 |
| Letter-number sequencing | 4.63 | 2.26 | 7.00 | 3.75 | 3.10 | 5.50 | 12.00 | .57 | 0.40 |
| Arithmetic | 1.75 | 1.04 | 5.69 | 3.00 | 2.16 | 8.13 | 9.50 | .28 | 0.67 |
| **Processing speed index** | 58.50 | 7.54 | 5.69 | 65.50 | 12.01 | 8.13 | 9.50 | .28 | 0.67 |
| Coding | 1.88 | 0.99 | 6.00 | 3.25 | 2.63 | 7.50 | 12.00 | .57 | 0.40 |
| Symbol search | 3.00 | 2.07 | 6.00 | 4.00 | 2.16 | 7.50 | 12.00 | .57 | 0.40 |
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| **Table 3S. Comparison of NEUROPSI Attention and Memory and ENI-2 visual memory subtest between atypical and typical WS group** | | | | | | | | | |
| **Variable** | **Typical group** | | | **Atypical group** | | | **Mann Whitney U** | | ***Cohen’s***  ***d*** |
| **M** | **SD** | **Mean rank** | **M** | **SD** | **Mean rank** | **U** | ***p*** |
| **Attention** | | | | | | | | | |
| Digits span | 4.00 | 3.16 | 6.06 | 5.25 | 3.50 | 7.38 | 12.50 | .57 | 0.35 |
| Cubes span | 2.88 | 2.36 | 6.06 | 3.50 | 3.11 | 7.38 | 12.50 | .57 | 0.35 |
| Visual detection | 7.00 | 3.93 | 7.13 | 5.00 | 4.24 | 5.25 | 11.00 | .46 | 0.51 |
| Verbal detection | 2.25 | 1.58 | 6.75 | 2.50 | 3.00 | 6.00 | 14.00 | .81 | 0.20 |
| **Verbal memory coding** | | | | | | | | | |
| Memory curve | 5.63 | 2.72 | 5.69 | 8.25 | 4.19 | 8.13 | 9.50 | .28 | 0.67 |
| Logical memory | 2.63 | 1.69 | 6.13 | 3.50 | 2.38 | 7.25 | 13.00 | .68 | 0.30 |
| Associated pairs | 6.63 | 4.50 | 7.63 | 2.50 | 1.29 | 4.25 | 7.00 | .15 | 0.98 |
| **Verbal memory evocation** | | | | | | | | | |
| Curve memory evocation | 4.50 | 2.00 | 6.44 | 4.75 | 3.86 | 6.63 | 15.50 | .93 | 0.05 |
| Curve memory with keys | 5.25 | 3.33 | 6.75 | 4.75 | 3.10 | 6.00 | 14.00 | .81 | 0.20 |
| Curve memory recognition | 6.00 | 5.01 | 6.69 | 4.50 | 2.65 | 6.13 | 14.50 | .81 | 0.15 |
| Logical memory evocation | 2.00 | 1.31 | 6.38 | 2.50 | 2.38 | 6.75 | 15.00 | .93 | 0.10 |
| Associated pairs evocation | 4.88 | 4.05 | 7.75 | 1.00 | 0.00 | 4.00 | 6.00 | .11 | 1.13 |
| **Visual memory coding** | | | | | | | | | |
| Learning figures | 1.88 | 1.46 | 6.69 | 1.75 | 1.50 | 6.13 | 14.50 | .81 | 0.15 |
| **Visual memory evocation** | | | | | | | | | |
| Learning figures evocation | 1.25 | 0.46 | 6.38 | 1.75 | 1.50 | 6.75 | 15.00 | .93 | 0.10 |
| Learning figures with keys | 1.88 | 1.64 | 6.44 | 2.00 | 2.00 | 6.63 | 15.50 | .93 | 0.05 |
| Learning figures recognition | 3.75 | 3.88 | 6.31 | 3.75 | 4.27 | 6.88 | 14.50 | .81 | 0.15 |
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| **Table 4S. Comparison of ENI-2 executive functions subtest between atypical and typical WS group** | | | | | | | | | |
| **Variable** | **Typical group** | | | **Atypical group** | | | **Mann Whitney U** | | ***Cohen’s***  ***d*** |
| **M** | **SD** | **Mean rank** | **M** | **SD** | **Mean rank** | **U** | ***p*** |
| **Verbal fluency** | | | | | | | | | |
| Verbal fluency (fruits) | 4.88 | 1.13 | 6.00 | 6.00 | 3.92 | 7.50 | 12.00 | .57 | 0.40 |
| Verbal fluency (animals) | 4.88 | 2.03 | 6.19 | 6.00 | 4.24 | 7.13 | 13.50 | .68 | 0.25 |
| Verbal fluency (phonological) | 4.50 | 2.27 | 5.69 | 7.00 | 3.56 | 8.13 | 9.50 | .28 | 0.67 |
| **Working Memory** | | | | | | | | | |
| Digits regression | 2.00 | 2.27 | 6.06 | 1.75 | 1.50 | 7.38 | 12.50 | .57 | 0.35 |
| Cubes regression | 2.63 | 1.76 | 6.06 | 1.75 | 1.50 | 7.38 | 12.50 | .57 | 0.35 |
| **Cognitive flexibility** | | | | | | | | | |
| Total trials | 7.13 | 0.64 | 7.13 | 6.75 | 0.50 | 5.25 | 11.00 | .46 | 0.51 |
| Total of correct responses | 7.63 | 3.46 | 7.44 | 4.25 | 4.57 | 4.63 | 8.50 | .21 | 0.79 |
| Percent of correct responses | 6.38 | 1.85 | 7.56 | 3.50 | 3.70 | 4.38 | 7.50 | .15 | 0.92 |
| Total of errors | 6.75 | 1.49 | 7.50 | 4.25 | 3.40 | 4.50 | 8.00 | .21 | 0.85 |
| Percent of errors | 6.88 | 1.81 | 7.44 | 4.50 | 3.87 | 4.63 | 8.50 | .21 | 0.79 |
| Total categories | 8.75 | 1.83 | 7.63 | 4.00 | 4.76 | 4.25 | 7.00 | .15 | 0.98 |
| Total perseverative responses | 5.25 | 3.20 | 7.13 | 3.50 | 5.00 | 5.25 | 11.00 | .46 | 0.51 |
| Percent perseverative responses | 5.25 | 3.28 | 7.13 | 3.75 | 5.50 | 5.25 | 11.00 | .46 | 0.51 |
|  | | | | | | | | | |

**Neuropsychology and behavioural results**

|  |  |  |  |  |  |  |  |  |  |  |  |  |
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| **Variable** |  |  |  |  |  |  |  |  |  |  |  |  |
| Subject | SW3 | SW5 | SW6 | SW8 | SW9 | SW10 | SW11 | SW13 | SW7 | SW2 | SW4 | SW12 |
| Genotype | + | + | + | + | + | + | + | + | - | - | - | - |
| Chronological age | 12 | 16 | 8 | 11 | 15 | 8 | 15 | 7 | 18 | 7 | 9 | 14 |
| Mental age | 6 | 8 | 4 | 5 | 6 | 5 | 7 | 4 | 12 | 5 | 4 | 6 |
| **WISC-IV/WAIS-IV\*^** | | | | | | | | | | | | |
| Full scale IQ | 50 | 47 | 48 | 46 | 40 | 56 | 49 | 54 | 65 | 68 | 47 | 41 |
| Verbal Comprehension Index | 50 | 57 | 57 | 50 | 50 | 63 | 69 | 50 | 72 | 77 | 63 | 50 |
| Perceptual Reasoning Index | 55 | 47 | 55 | 61 | 45 | 61 | 45 | 65 | 66 | 63 | 51 | 45 |
| Working Memory Index | 68 | 68 | 59 | 59 | 50 | 65 | 62 | 74 | 52 | 80 | 59 | 50 |
| Processing Speed Index | 68 | 56 | 56 | 50 | 50 | 70 | 56 | 62 | 75 | 75 | 50 | 62 |
| Similarities | 3 | 5 | 5 | 1 | 3 | 6 | 5 | 1 | 4 | 8 | 6 | 3 |
| Vocabulary | 1 | 2 | 2 | 3 | 1 | 4 | 3 | 3 | 4 | 7 | 4 | 1 |
| Comprehension | 1 | 1 | 1 | 1 | 1 | 1 | 6 | 1 | 2 | 3 | 1 | 1 |
| Information | 4 | 1 | 2 | 2 | 1 | 3 | 5 | 4 | 5 | 6 | 1 | 1 |
| Tracks | 4 | 4 | 2 | 5 | 5 | 5 | 4 | 1 |  | 4 | 4 | 1 |
| Block design | 2 | 1 | 1 | 2 | 1 | 3 | 1 | 5 | 3 | 1 | 1 | 1 |
| Picture concepts | 1 | 1 | 1 | 5 | 1 | 2 | 1 | 2 | 1 | 5 | 1 | 1 |
| Matrix reasoning | 5 | 2 | 6 | 4 | 1 | 6 | 1 | 6 | 5 | 6 | 4 | 1 |
| Picture completion | 1 | 3 | 5 | 1 | 1 | 2 | 1 | 3 | 4 | 5 | 2 | 1 |
| Digit span | 4 | 6 | 1 | 1 | 1 | 1 | 4 | 3 | 3 | 5 | 2 | 1 |
| Letter-number sequencing | 5 | 3 | 5 | 5 | 1 | 7 | 3 | 8 | 2 | 8 | 4 | 1 |
| Arithmetic | 2 | 1 | 2 | 1 | 1 | 4 | 1 | 2 | 1 | 6 | 3 | 2 |
| Coding | 3 | 1 | 2 | 1 | 1 | 3 | 3 | 1 | 6 | 5 | 1 | 1 |
| Symbol search | 5 | 3 | 2 | 1 | 1 | 6 | 1 | 5 | 4 | 6 | 1 | 5 |
| **NEUROPSI Attention and Memory^** | | | | | | | | | | | | |
| Digit span | 2 | 7 | 1 | 5 | 1 | 6 | 1 | 9 | 7 | 9 | 1 | 4 |
| Cubes span | 1 | 6 | 1 | 6 | 1 | 5 | 2 | 1 | 3 | 8 | 1 | 2 |
| Visual detection | 12 | 8 | 3 | 6 | 1 | 11 | 10 | 5 | 1 | 7 | 10 | 2 |
| Verbal detection | 5 | 2 | 1 | 1 | 1 | 3 | 1 | 4 | 1 | 7 | 1 | 1 |
| Digits regression | 4 | 7 | 1 | 1 | 1 | 1 | 4 | 1 | 4 | 1 | 1 | 1 |
| Cubes regression | 1 | 3 | 2 | 1 | 3 | 4 | 6 | 1 | 1 | 4 | 1 | 1 |
| Memory curve | 3 | 10 | 4 | 4 | 5 | 3 | 9 | 7 | 8 | 14 | 4 | 7 |
| Logical memory | 1 | 3 | 2 | 2 | 2 | 4 | 1 | 6 | 1 | 6 | 5 | 2 |
| Associated pairs | 1 | 1 | 6 | 5 | 14 | 11 | 8 | 7 | 3 | 1 | 2 | 4 |
| Face learning | 1 | 1 | 3 | 5 | 3 | 9 | 3 | 9 | 3 | 7 | 6 | 1 |
| Curve memory evocation | 4 | 5 | 3 | 1 | 7 | 4 | 7 | 5 | 7 | 2 | 1 | 9 |
| Curve memory with keys | 6 | 10 | 4 | 2 | 5 | 1 | 10 | 4 | 5 | 9 | 2 | 3 |
| Curve memory recognition | 4 | 12 | 1 | 12 | 6 | 1 | 11 | 1 | 1 | 6 | 7 | 4 |
| Logical memory evocation | 2 | 1 | 1 | 2 | 1 | 4 | 1 | 4 | 1 | 6 | 2 | 1 |
| Associated pairs evocation | 1 | 1 | 3 | 1 | 8 | 12 | 7 | 6 | 1 | 1 | 1 | 1 |
| **ENI-2^** | | | | | | | | | | | | |
| Learning figures | 2 | 1 | 5 | 3 | 1 | 1 | 1 | 1 | 1 | 4 | 1 | 1 |
| Learning figures evocation | 1 | 2 | 1 | 1 | 1 | 2 | 1 | 1 | 1 | 4 | 1 | 1 |
| Learning figures with keys | 1 | 1 | 4 | 1 | 1 | 5 | 1 | 1 | 1 | 5 | 1 | 1 |
| Learning figures recognition | 7 | 8 | 1 | 1 | 1 | 1 | 10 | 1 | 1 | 10 | 3 | 1 |
| Verbal fluency (fruits) | 3 | 4 | 6 | 5 | 4 | 6 | 6 | 5 | 1 | 10 | 5 | 8 |
| Verbal fluency (animals) | 3 | 1 | 7 | 5 | 5 | 6 | 5 | 7 | 1 | 10 | 4 | 9 |
| Verbal fluency (phonological) | 1 | 4 | 6 | 4 | 6 | 8 | 2 | 5 | 4 | 12 | 5 | 7 |
| Total trials | 7 | 6 | 7 | 7 | 7 | 8 | 7 | 8 | 6 | 7 | 7 | 7 |
| Correct responses (Total) | 9 | 10 | 4 | 2 | 13 | 9 | 7 | 7 | 3 | 2 | 11 | 1 |
| Correct responses (%) | 7 | 7 | 4 | 4 | 9 | 8 | 5 | 7 | 2 | 2 | 9 | 1 |
| Errors (Total) | 7 | 7 | 6 | 4 | 9 | 8 | 6 | 7 | 3 | 4 | 9 | 1 |
| Errors (%) | 8 | 7 | 5 | 4 | 9 | 9 | 6 | 7 | 3 | 4 | 10 | 1 |
| Categories (Total) | 10 | 9 | 7 | 6 | 9 | 12 | 9 | 8 | 1 | 3 | 11 | 1 |
| Failure to maintain set | 11 | 11 | 11 | 11 | 5 | 12 | 11 | 12 | 5 | 11 | 5 | 11 |
| Perseverative responses (Total) | 6 | 7 | 1 | 1 | 8 | 10 | 4 | 5 | 1 | 1 | 11 | 1 |
| Perseverative responses (%) | 6 | 6 | 1 | 1 | 9 | 10 | 4 | 5 | 1 | 1 | 12 | 1 |
| **DTVP-3^\*** | | | | | | | | | | | | |
| Eye-hand coordination | 6 | 1 | 1 | NA | NA | 2 | 2 | 1 | 1 | 1 | 1 | 6 |
| Copying | 7 | 4 | 1 | NA | NA | 6 | 7 | 1 | 6 | 2 | 1 | 6 |
| Figure-ground | 9 | 11 | 7 | NA | NA | 9 | 7 | 6 | 6 | 7 | 7 | 11 |
| Visual closure | 5 | 3 | 3 | NA | NA | 4 | 3 | 3 | 3 | 1 | 2 | 2 |
| Form constancy | 11 | 9 | 10 | NA | NA | 12 | 11 | 12 | 8 | 3 | 11 | 8 |
| Visual-motor integration index | 79 | 55 | 45 | NA | NA | 64 | 67 | 45 | 61 | 48 | 45 | 76 |
| Reduced motor response index | 90 | 86 | 80 | NA | NA | 90 | 82 | 82 | 73 | 60 | 80 | 82 |
| General visual perception index | 85 | 73 | 66 | NA | NA | 79 | 75 | 67 | 68 | 55 | 66 | 79 |
|  |  |  |  |  |  |  |  |  |  |  |  |  |
| **Variable** |  |  |  |  |  |  |  |  |  |  |  |  |
| **Subject** | SW3 | SW5 | SW6 | SW8 | SW9 | SW10 | SW11 | SW13 | SW7 | SW2 | SW4 | SW12 |
| **SENAϮ** | | | | | | | | | | | | |
| Global problems index | 54 | 41 | 85 | 72 | 49 | 50 | 34 | 51 | 50 | 50 | 60 | 52 |
| Emotional problems index | 60 | 41 | 78 | 71 | 46 | 46 | 36 | 39 | 49 | 44 | 56 | 48 |
| Behavioural problems index | 47 | 42 | 83 | 61 | 49 | 51 | 38 | 62 | 45 | 53 | 58 | 55 |
| executive functions index | 51 | 45 | 76 | 70 | 56 | 54 | 40 | 54 | 59 | 53 | 62 | 55 |
| Personal sources index | 50 | 56 | 24 | 31 | 55 | 41 | 51 | 47 | 33 | 34 | 48 | 30 |
| Depression | 52 | 44 | 108 | 71 | 47 | 49 | 37 | 49 | 47 | 49 | 63 | 48 |
| Anxiety | 71 | 43 | 82 | 74 | 50 | 55 | 39 | 44 | 56 | 53 | 53 | 48 |
| Social anxiety | 57 | 39 | 41 | 57 | 37 | 37 | 35 | 33 | 54 | 41 | 46 | 41 |
| Somatic complaints | 51 | 45 | 53 | 66 | 55 | 47 | 47 | 40 | 41 | 40 | 56 | 56 |
| Attention problems | 51 | 44 | 70 | 70 | 59 | 61 | 49 | 63 | 62 | 57 | 68 | 59 |
| Hyperactivity | 48 | 54 | 63 | 68 | 49 | 52 | 41 | 55 | 60 | 54 | 53 | 56 |
| Anger management problems | 57 | 45 | 94 | 61 | 47 | 54 | 37 | 57 | 42 | 40 | 54 | 62 |
| Aggression | 43 | 43 | 78 | 57 | 49 | 44 | 41 | 63 | 46 | 63 | 63 | 46 |
| Disruptive behaviour | 41 | 42 | 62 | 60 | 48 | 54 | 38 | 61 | 52 | 54 | 54 | 55 |
| Unusual behaviour | 46 | 64 | 85 | 79 | 62 | 71 | 43 | 56 | 47 | 76 | 100 | 53 |
| Emotional regulation problems | 54 | 41 | 82 | 70 | 50 | 58 | 41 | 54 | 45 | 52 | 63 | 54 |
| Rigidity | 50 | 45 | 63 | 57 | 60 | 42 | 37 | 39 | 62 | 47 | 53 | 46 |
| Isolation | 64 | 43 | 83 | 76 | 69 | 54 | 42 | 47 | 96 | 77 | 71 | 79 |
| Social integration | 38 | 46 | 26 | 43 | 48 | 58 | 49 | 58 | 36 | 38 | 52 | 29 |
| Emotional intelligence | 52 | 59 | 47 | 40 | 66 | 42 | 54 | 63 | 41 | 35 | 36 | 33 |
| Disposition to the study | 61 | 59 | 19 | 22 | 47 | 30 | 50 | 22 | 33 | 40 | 47 | 42 |
| **ABAS-II^\*** | | | | | | | | | | | | |
| Communication | 1 | 8 | 4 | 1 | 1 | 5 | 2 | 9 | 3 | 5 | 2 | 1 |
| Academic skills | 1 | 1 | 1 | 1 | 1 | 1 | 1 | 1 | 1 | 3 | 1 | 1 |
| Self-direction | 9 | 13 | 1 | 6 | 4 | 8 | 4 | 7 | 1 | 4 | 6 | 1 |
| Leisure | 5 | 9 | 3 | 5 | 10 | 9 | 3 | 10 | 1 | 6 | 1 | 1 |
| Social | 11 | 12 | 7 | 6 | 10 | 9 | 7 | 9 | 6 | 7 | 6 | 1 |
| Use of community resources | 3 | 3 | 3 | 1 | 1 | 11 | 1 | 8 | 1 | 7 | 5 | 1 |
| Home life | 9 | 8 | 11 | 2 | 8 | 7 | 4 | 11 | 1 | 7 | 10 | 8 |
| Health and security | 10 | 5 | 3 | 1 | 8 | 11 | 3 | 7 | 1 | 5 | 6 | 1 |
| Self-care | 4 | 5 | 2 | 1 | 3 | 5 | 6 | 10 | 3 | 2 | 4 | 1 |
| Conceptual index | 66 | 84 | 58 | 60 | 58 | 71 | 60 | 76 | 54 | 67 | 63 | 54 |
| Social index | 88 | 98 | 75 | 77 | 96 | 92 | 75 | 94 | 68 | 82 | 68 | 51 |
| Practical index | 77 | 71 | 69 | 51 | 70 | 89 | 61 | 91 | 53 | 71 | 67 | 58 |
| General adaptive coefficient | 74 | 81 | 63 | 57 | 70 | 83 | 59 | 86 | 56 | 70 | 67 | 53 |
| *Notes:* ***+=****With GTF2IRD2,* ***-=****Loss GTF2IRD2; ^=Scale scores; \*=Index scores;* Ϯ*=T scores, NA=Not Applicate* | | | | | | | | | | | | |