**Table S1 Quantification of attention deficits, social interaction and cognition of the NSDHL-containing patient**

**­­­­­­­ Subject Patient**

**Swan-son Nolan and Pelham, Version IV (SNAP-IV) Aged 7years 1month**

Inattention 14/1.6

Hyperactivity/impulsivity 11/1.2

Combined type 25/1.4

Oppositional Items 17/2.1

**Autism Diagnostic Observation Schedule (ADOS) M3 Aged 8years 4 months**

Communication score (cutoff score for autism is ≥3) 4

Social interaction score (cutoff score for autism is ≥6) 6

Combined ADOS score (cutoff score for autism is ≥10) 10

Play +Stereotypic behavior and restricted interests 1+2

**Autism Diagnostic Interview-Revised (ADI-R) Aged 8years 4 months**

Qualitative Abnormalities in Reciprocal Social Interaction 16

(cutoff score=10)

Qualitative Abnormalities in Communication 12, 7

(cutoff score for verbal &nonverbal =8,7 )

Restricted Repetitive and Stereotyped Patterns of Behavior 2

(cutoff score=3)

Abnormality of Development Evident at or Before 36 Months 3

(cutoff score=1)

**Wechsler Intelligence Scale for Children (WISC) Aged 9years 8 months**

IQS of Verbal Scale 96

IQS of Performance Scales 90

Full Scale IQ 92

**Table S2 Primers used for NSDHL variant in the study**

|  |  |  |
| --- | --- | --- |
| **Primer for** | **Primer** | **Primer sequence** |
| **DNA** | Exon Forward Primer | CTTTCTCCCACTCATTCCTCAG |
| Exon Reverse Primer | CCCACCCTATTCCCAACTTATT |
| **cDNA** | Exon Forward Primer | CGCATGGCTGGCACATTCC |
| Exon Reverse Primer | ACGGTCCTCTCCATAGCATCATCC |

**Table S3-1 Tissue expression of genes in the duplicated region in Gene**

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| *ZNF 185* | *NSDHL* | *MAGEA 12* | *MAGEA 6* | | *MAGEA3* | | | *MAGEA2* | *CSAG4* | *CSAG3* | *CSAG2* | *CSAG1* | *CETN2* | **tissue expression of Gene**  **(Mean PRKM)**  **Gene in the**  **duplicated region** |
| 5.316±1.535 | 13.963±1.993 | - | | - | | - | - | | - | - | - | - | 30.719±3.827 | adrenal |
| 2.608±0.477 | 5.066±0.965 | - | | - | | - | - | | - | 0.079±0.068 | 0.061±0.046 | 0.076±0.08 | 15.788±2.418 | appendix |
| 2.602±0.953 | 2.772±0.441 | - | | - | | - | - | | - | - | - | - | 12.308±2.275 | bone marrow |
| **0.978±0.192** | **10.107±1.253** | **0.094±0.027** | | **-** | | **-** | **-** | | **-** | **0.054±0.064** | **0.058±0.042** | **0.168±0.039** | **28.291±1.274** | **brain** |
| 1.911±0.908 | 6.173±1.842 | - | | - | | - | - | | - | - | - | - | 15.491±2.785 | colon |
| - | 6.51±2.448 | - | | - | | - | - | | - | - | - | - | 9.666±0.212 | duodenum |
| 14.04±12.282 | 6.375±1.779 | - | | - | | - | - | | - | - | - | - | 24.998±4.936 | endometrium |
| 112.2±25.541 | 14.3380.733 | - | | - | | - | - | | - | - | - | - | 24.998±4.936 | esophagus |
| 1.518±0.134 | 11.563±2.395 | 0.082±0.043 | | - | | 0.191±0.117 | - | | - | 0.174±0.124 | 0.074±0.05 | 0.083±0.117 | 22.985±2.553 | fat |
| 3.525±0.506 | 4.689±1.02 | - | | - | | - | - | | - | - | - | - | 25.494±5.526 | gall bladder |
| - | 5.634±0.834 | - | | - | | - | - | | - | - | - | - | 16.45±2.033 | heart |
| 4.002±0.946 | 0.886±0.667 | - | | - | | - | - | | - | 0.085±0.074 | - | - | 38.151±2.829 | kidney |
| - | 9.555±1.896 | - | | - | | - | - | | - | - | - | - | 10.856±2.829 | liver |
| 5.595±0.918 | 7.142±0.907 | - | | - | | - | - | | - | 0.068±0.072 | - | - | 30.61±14.161 | lung |
| 1.679±0.254 | 4.113±0.895 | - | | - | | - | - | | - | 1.14±0.314 | 0766±0.214 | 0.231±0.136 | 10.083±1.654 | lymph node |
| - | 4.518±0.265 | - | | - | | - | - | | - | - | - | - | 33.279±0.951 | ovary |
| - | 1.083±0.109 | - | | - | | - | - | | - | - | - | - | 3.421±0.245 | pancreas |
| 11.236±2.505 | 5.095±1.406 | - | | - | | - | 0.068±0.065 | | - | - | - | - | 13.296±1.786 | placenta |
| 9.868±1.736 | 7.48±1.123 | - | | - | | - | - | | - | - | - | - | 37.352±6.814 | prostate |
| 1.689±0.344 | 2.048±0.586 | - | | - | | - | - | | - | - | - | - | 6.492±2.152 | salivary gland |
| 20.817±6.923 | 6.393±1.201 | - | | - | | - | - | | - | 0.652±0.08 | 0.427±0.089 | - | 9.601±2.085 | skin |
| - | 5.053±0.853 | - | | - | | - | - | | - | - | - | - | 11.274±1.878 | small intestine |
| 1.66±0.295 | 4.059±0.339 | - | | - | | - | - | | - | 0.811±0.448 | 0.523±0.246 | - | 10.475±0.769 | spleen |
| 3.101±1.401 | 5.089±0.638 | - | | - | | - | - | | - | - | - | - | 11.416±2.446 | stomach |
| 1.786±0.386 | 6.063±1.523 | 1.817±0.451 | | - | | 4.808±1.152 | 2.763±0.804 | | 0.261±0.131 | 1.559±0.379 | 10.25±0.233 | 0.641±0.248 | 20.494±4.775 | testis |
| 9.735±4.351 | 7.312±1.088 | - | | - | | - | - | | - | - | - | 0.070±0.094 | 47.158±15.976 | thyroid |
| 3.491±0.764 | 5.074±0.464 | - | | - | | - | - | | - | - | - | - | 23.359±3.508 | urinary bladder |

**-:tiny or no expression**

**Table S3-2 Brain expression from Allen Brian and known related diseases in the OMIM of genes in the duplicated region**

|  |  |  |
| --- | --- | --- |
| **Genes in the**  **duplicated region** | **Brain expression from Allen Brian Atlas-Developmental Transcriptome**  **（average in log2 RPKM）** | **Known related diseases in OMIM** |
| *CETN2* | 4.678 | NR |
| *CSAG1* | 0.324 | NR |
| *CSAG2* | 0.001 | N/A |
| *CSAG3* | 0.001 | N/A |
| *CSAG4* | 0.018 | N/A |
| *MAGEA2* | 0.000 | NR |
| *MAGEA3* | 0.001 | NR |
| *MAGEA6* | 0.002 | Somatic Mutation in  Pancreatic Cancer |
| *MAGEA 12* | 0.109 | NR |
| *NSDHL* | 3.710 | CHILD syndrome  CK syndrome |
| *ZNF 185* | 1.274 | downregulation may be of biologic relevance in lung carcinogenesis |

**N/A: not available NR: not been reported**