**Social and emotional characteristics of girls and young women with DDX3X-associated intellectual disability: A descriptive and comparative study**

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**Supplementary Material**

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**Supplementary Information 1. Genetic diagnoses in the ID-comparison group**

|  |  |
| --- | --- |
| **Gene** | **N** |
| *ARID1B* | 3 |
| *CASK* | 1 |
| *CTNNB1* | 1 |
| *DYRK1A* | 1 |
| *EHMT1* | 3 |
| *GRIN2A* | 1 |
| *KAT6B* | 1 |
| *SETD5* | 4 |
| *SHANK1* | 2 |
| *STXBP1* | 5 |
| *TRIO* | 1 |
| **Total** | 23 |

**Supplementary Information 2. Participants’ gene variant information**

| **Participant no.** | **Decipher/100K Genome ID** | **Inheritance** | **Variant type** | **Amino acid change** | **Nucleotide change** | **Pathogenicity** | **ID severity** |
| --- | --- | --- | --- | --- | --- | --- | --- |
| 1 | 272869 | De novo | Missense | p.Cys467Tyr | c.1400G>A | Likely pathogenic | Severe |
| 2 | 273466 | De novo | Splice (acceptor) | - | c.46-2A​>G | Likely pathogenic | Mild |
| 3 | 272597 | De novo | Stop-gained | p.Ser24Ter | c.71C​>A | Pathogenic | Mild |
| 4 | 264193 | De novo | Stop-gained | p.Arg291Ter | c.871C​>T | Likely pathogenic | Mild |
| 5 | 261890 | Inherited | Missense | p.Arg533His | c.1598G​>A | Likely pathogenic | Moderate |
| 6 | 267102 | De novo | Missense | p.Ala392Pro | c.1174G​>C | Likely pathogenic | Severe |
| 7 | 267105 | De novo | Missense | p.Ala392Pro | c.1174G​>C | Likely pathogenic | Moderate |
| 8 | 261162 | De novo | Stop-gained | p.Arg45Ter | c.133C​>T | Pathogenic | Moderate |
| 9 | 290561 | De novo | Splice (donor) | - | c.281+1G​>C | Likely pathogenic | Moderate |
| 10 | 300283 | De novo | Missense | p.Arg474Cys | c.1420C​>T | Likely pathogenic | Mild |
| 11 | - | - | - | - | - | - | Moderate |
| 12 | 277307 | De novo | Splice (donor) | - | c.45+1G>T | Pathogenic | Mild |
| 13 | 303778 | De novo | Frameshift | p.Lys208SerfsTer13 | c.623delA | Pathogenic | Borderline |
| 14 | - | De novo | Missense | Cys341Arg | c.1021T>C | - | Severe |
| 15 | 282969 | De novo | Splice donor | - | c.45+1G>A | Pathogenic | Profound |
| 16 | - | - | - | - | - | - | Moderate |
| 17 | - | De novo | Splice (acceptor) |  | c.766-8\_766-2del | Likely pathogenic | Mild |
| 18 | - | - | - | - | - | - | Moderate |
| 19 | - | Unknown | Frameshift | p.Asn124LysfsTer5 | c.372\_373delCA | - | Mild |
| 20 | 307552 | De novo | Frameshift | p.Phe83Ter | c.241\_242insCTAG | Pathogenic | Borderline |
| 21 | 210015169 | De novo | Frameshift | p.Gly153Leufs\*69 | c.455\_456dupCT | Pathogenic | Mild |
| 22 | 281511 | De novo | Missense | p.Ser542Leu | c.1625C>T | Likely pathogenic | Moderate |
| 23 | - | Unknown | Splice site | - | c.765+1G>A | Pathogenic | Mild |

**Supplementary Information 3. ADI score profiles - Current**

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*Note.* Orange items = Language and Communication, Blue = Social Development and Play, Green = Interests and Behaviours, Yellow = General Behaviours.

**Supplementary Information 4. ADI score profiles – Most Abnormal / age 5**



*Note.* Orange items = Language and Communication, Blue = Social Development and Play, Green = Interests and Behaviours, Yellow = General Behaviours.

**Supplementary Information 5. Scatterplots for within-group correlations**



**Supplementary Information 6. Regression Models with Group and Interaction Terms**

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Dependent variable**Predictors | **ANOVA** | **Adjusted R2** | **B** | **Std. Err** | **Beta (β)** | **t** | **p** | **95% CI for B** |
| **Anxiety (n = 44)** | F (3,40) = 2.25, p =.10 | .14 |  |  |  |  |  |  |
| Group |  |  | -20.96 | 14.71 | -.90 | -1.41 | .17 | [-5.45, 9.02] |
| Adaptive ability |  |  | -.63 | .39 | -.72 | -1.60 | .12 | [-1.42, .17] |
| Group\* Adaptive ability |  |  | .29 | .26 | .89 | 1.11 | .27 | [-.24, .83] |
| **Self-Injury (n = 44)** | F (3,40) = 6.68, p =.001 | .28 |  |  |  |  |  |  |
| Group  |  |  | -5.13 | 2.28 | -1.26 | -2.25 | .03 | [-9.74, -.51] |
| Adaptive ability |  |  | -.16 | .06 | -1.05 | -2.66 | .01 | [-.29, -.04] |
| Group\*Adaptive ability |  |  | .07 | .04 | 1.24 | 1.74 | .09 | [-.01, .15] |
| **Self-Injury (n = 44)** | F (3,40) = 7.39, p <.001 |  |  |  |  |  |  |  |
| Group |  |  | 5.62 | 2.55 | 1.38 | 2.20 | .03 | [.46, 10.78] |
| Anxiety |  |  | .26 | .08 | 1.49 | 3.35 | .002 | [.10, .42] |
| Group\*Anxiety |  |  | -.13 | .05 | -1.81 | -2.69 | .01 | [-.22, -.03] |
| **Self-Injury (n = 42)** | F (3,38) = 4.85, p =.006 |  |  |  |  |  |  |  |
| Group |  |  | 4.76 | 3.16 | 1.17 | 1.51 | .14 | [-1.64, 11.16] |
| Autism char. |  |  | .17 | .07 | 1.14 | 2.55 | .02 | [.03, .30] |
| Autism char.\*Self Injury |  |  | -.08 | .04 | -1.69 | -1.94 | .06 | [-.16, .00] |
| *Note*. Method = Enter. |