|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
|  |  | **GDD** | **ID** | **Speech delay** | **Autism** | **Feeding problems** | **Behaviour** | **Microcephaly** | **Cranio-facial features** | **Short stature** | **Seizures** | **FTT** | **Hypotonia** | ***ASXL3* variant (Amino acid change)** |
| Bainbridge et al |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
|  | p1 | + | + |  |  | + |  | + | Arched eyebrows; posteriorly rotated ears; anteverted nares; large fontanels; retrognathia; hirsutism. | + |  | + | + | Gln404Ter |
|  | p2 | + | + | + |  | + |  | + | Arched eyebrows; low posteriorly rotated ears; anteverted nares; hypertelorism; short nose | + |  | + | + | Gln404Ter |
|  | p3 | + | + | + |  | - |  |  | Prominent forehead; short anteverted nares. |  |  | + | + | Thr659fsTer41 |
|  | p4 | + | + | + |  | + |  |  | Prominent forehead; arched eyebrows; hypoplastic nose; low set ears; hirsutism | + |  | + | Variable tone | Pro474fs |
| Dinwiddie et al |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
|  | p1 | + | + | + | + | + | Poor sleep; aggressive behaviour | + | Trigonocephaly with metopic ridging; epicanthic folds; hypoplastic nose; wide and posteriorly rotated ears; myopia; hypertrichosis | + | - | + | + | Gln633ValfsTer13 |
| Srivastava et al |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
|  | p1 | + | + | + |  | + |  | - |  |  | + | + | + | Thr484NfsTer5 |
|  | p2 | + | + | + |  | + | Aggressive behaviour | - | Anteverted nares; small chin; downslanting palpebral fissures |  | - | + | + | Arg1444Ter |
|  | p3 | + |  | + |  | + |  | - | Kyloglossia (frenulotomy); arched eyebrows |  | - | + | + | Gln112Ter |
| Hori et al |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
|  | p1 | + | + | + | + | + | Poor sleep |  | Prominent forehead; thick eyebrows; downslanting palpebral fissures; strabismus; depressed nasal bridge; hirsutism | + | - | + | - | Arg1729Cys |
| Balasubramanian et al |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
|  | p1 | + | + | + | + | - | Autistic traits |  | Downslanting palpebral fissures; long nose; thin upper lip; high arched palate |  | - | - | + | Arg1444Ter |
|  | p2 | + | + | + | + | + | Autistic traits |  | Hypertelorism; hirsutism; thin eyebrows; high arched palate; squint | + | + | + | + | Ala401GlnfsTer8 |
|  | p3 | + | + | + | - | - | Normal |  | Hypertelorism; downslanting palpebral fissures; narrow high arched palate; strabismus |  | - | - | + | Tyr358Ter |
|  | p4 | p1 | + | + | + | + | Autistic traits |  | Thick eyebrows; prominent forehead | + | - | + | + | Gln1382Ter |
|  | p5 | + | + | + | + | + | Normal |  | Posteriorly rotated ears; dental overcrowding; high arched palate; strabismus | - | - | - | + | Gln595Ter |
|  | p6 | + | + | + | - | + | Agitation |  | Tongue tie; micrognathia; high arched palate; strabismus | - | - | - | + | His1119ProfsTer7 |
|  | p7 | + | + | + | + | - | Poor sleep |  | Synophrys; micrognathia; downslanting palpebral fissures | - | - | - | + | Leu362AlafsTer23 |
|  | p8 | + | + | + | + | + | Tendency to get frustrated |  | Prominent nasal bridge; mild synophrys; crowded teeth; high arched palate; bifid uvula; bilateral strabismus | + | + | + | + | Leu1212Ter |
|  | p9 | + | + | + | + | + |  |  | Strabismus | - | - | + | + | Gly1045ValfsTer |
|  | p10 | + |  | + | + | + | Sleep disorder |  | Upslanting palpebral fissures; twin hair crowns; high arched palate | - | - | - | + | Arg1060ProfsTer50 |
|  | p11 | + | + |  | + | + | Grunting; bruxism |  | Downslanting palpebral fissures; high arched palate | - | + | - | + | Asp497Ter |
|  | p12 | + | + | + | + | + |  |  | Downslanting palpebral fissures; high arched palate; strabismus | - | + | - | + | Asn498Ter |
| Chinen et al |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
|  | p1 | + | + | + |  |  |  |  | Prominent forehead; hypertelorism; short nose; high arched eyebrows with mild synophrys; long philtrum | + | - | + | + | Pro1010LeufsTer14 |
| Kuechler et al |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
|  | p1 | + | + | + |  | + |  | + | Long face; prominent forehead; arched eyebrows; downslanting palpebral fissures; small and anteverted nares | + | - | + | + | Ser407AlafsTer2 |
|  | p2 | + | + | + |  | + | Poor eye contact; open mouth appearance | - | Long face; prominent forehead; arched eyebrows; downslanting palpebral fissures; cleft palate; bifid uvula; depressed nasal bridge; anteverted nares | - | - | - | + | Glu457Ter |
|  | p3 | + | + | + |  | + |  | - | Long face; prominent forehead; high-arched palate; arched eyebrows; downslanting palpebral fissures; strabismus; anteverted nares | - | - | - | + | Arg1036Ter |
|  | p4 | + | + | + |  | + | Poor eye contact | - | Long face; prominent forehead; high-arched palate; arched eyebrows; downslanting palpebral fissures; strabismus; anteverted nares | - | - | - | + | Cys1165Ter |
|  | p5 | + | + | + |  | + | OCD | - | Long face; high-arched palate; arched eyebrows; downslanting palpebral fissures; strabismus; anteverted nares; | - | + | + | + | Glu1205Ter |
|  | p6 | + | + | + |  | + | Stereotypy | - | Long face; prominent forehead; high-arched palate; arched eyebrows; downslanting palpebral fissures; anteverted nares; strabismus | - | + | + | + | Val1358LeufsTer8 |
| Contreas-Capetillo et al |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
|  | p1 | + | + | + |  | + |  | + | Prominent forehead; hypertelorism; thin eyebrows (mild synophrys); anteverted nares with hypoplastic alae nasi; high arched palate; low set ears | + | + | + | + | Glu998fs |
| Koboldt et al (siblings) |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
|  | p1 | + | + | + | - | + | Autism |  | Long face; prominent forehead; downslanting palpebral fissures; hypoplastic alae nasi; high narrow palate; micrognathia; hirsutism | + | + | + | + | Arg1036Ter |
|  | p2 | + | + | + | + | + | Autism |  | Long face; prominent forehead; downslanting palpebral fissures; hypoplastic alae nasi; micrognathia; hirsutism | + | + | + | + |  |
| Verhoeven et al |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
|  | p1 | + | + | + | + | - | Aggressive behaviour; self-harming; OCD | + | Deep set eyes; prominent nose |  | + |  |  | Ser2238fs |
| Barcot et al |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
|  | p1 |  |  |  |  |  |  |  | High arched palate; low set posteriorly rotated ears; hypertrichosis; arched eyebrows |  |  |  |  | Glu440GlyfsTer7 |
| Qiao et al |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
|  | p1 | + |  |  |  | - | No eye contact; hypersomnia | + | Hirsute prominent forehead; long face; arched eyebrows; flat nose bridge | + | - | + | + | Ser1155Ter |
| Wayhelova et al |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
|  | p1 | + | + | + | + | + | Autism | + | Palate malformations; strabismus |  | - |  | + | Arg1004GlufsTer21 |
| Myer et al |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
|  | p1 | + | + | + | - |  |  |  | Periorbital fullness; full cheeks; open mouth with drooling, thick lower vermillion, small ears |  | + |  | + | Arg1036Ter |
|  | p2 | + | + | + |  | + |  | + |  | + | + | + |  | c.3039+1G >A; p. ?? |
|  | p3 | + | + |  | + |  | Autism | + |  | - | + | - |  | Thr1106ArgfsTer36 |
| Yu et al |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
|  | p1 | + | + | + | - | + |  | + | Arched eyebrows; anteverted nares; everted lower lip; reduced facial expression; hypertelorism |  | - | + | + | Lys1103Ter |
|  | p2 | + | + | + | + | + | Autism | + | Prominent forehead; arched eyebrows; anteverted nares; reduced facial expression; hypertelorism |  | - | - | + | Gln1819Ter |
|  | p3 | + | + | + | - | + |  | + | Prominent forehead; arched eyebrows; anteverted nares; hypertelorism |  | - | + | + | Glu459fsTer |
|  | p4 | + | + | + | - | + |  | - | Prominent forehead; downslanting palpebral fissures; hypertelorism |  | - | - | + | Arg1467Ter |

GDD: global developmental delay, ID: intellectual disability, FTT: failure to thrive, OCD: obsessive-compulsive disorder, +: present, -: absent,

**Supplementary Table 1: Characteristics of previously reported patients with Bainbridge-Ropers syndrome**

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