e-Table-1: Individual data on growth, development and clinical examination of 45 patients with *FOXG1* mutations/deletions.

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| *Patient/ gender* | Age at last follow-up | Mutation  | First concerns (Age / Nature) | OFC at birth (SD score)  | OFC posnatal (SD score)  | Motor and speech development  | Behavior and sleep disturbances | Feeding difficulties | Clinical examination | Eye examination  |
| *Tel02/M* | 4 y 6 m | p.Gln73dup | 5 m/ DD | 33,5 cm (-0,7 SD) | 44,5 cm (-4,4 SD) | Severe DD and ID, no speech | Impaired sleep pattern, inappropriate laughing and crying, screaming spells | Yes, since birth | Axial hypotonia with limb spasticity, orofacial dyskinesia and limb dystonia | No strabismus, inconsistent eye contact  |
| *Trs1/F* | 3 y 6 m | p.Gln86Profs\*35 | 3 m/ Axial hypotonia, esotropia | 33,5 cm (-0,7 SD) | 43 cm (-4,8 SD) | Severe DD and ID, no speech | Impaired sleep pattern, inappropriate laughing and crying, screaming spells | Absent | Axial hypotonia with limb spasticity, permanent limb dyskinesia, scoliosis | Strabismus |
| *Im11/F* | 1 y 7 m | p.Gln86Profs\*35 | 2 m/ Esotropia with poor visual following | 33 cm (-1,7 SD) | 42 cm (-4,1 SD) | Severe DD and ID, no speech | Impaired sleep pattern | Yes, since birth | Axial hypotonia with limb spasticity, dyskinesia, dystonia, choreoathetosis, round-faced,  | Bilateral esotropia |
| *Im05/M* | 10 y 2 m | p.Gln86Profs\*35 | 4 m/ Weeping, agitation, opisthotonos | 34 cm (-0,7 SD) | 46 cm (-5,4 SD) | Severe DD and ID, no speech | Impaired sleep pattern, inappropriate laughing and crying, screaming spells | Yes, percutaneous gastrostomy | Axial hypotonia with limb spasticity, hyperkinetic movement, dystonia, orofacial dyskinesia, stereotypical movements, scoliosis | Strabismus, intermittent eye contact and smiling intentionally |
| *Nan02/F* | 3 y | p.Gln86Aspfs\*34 | 20 m/ Seizures | 34 cm (0 SD) | 43 cm (-4,5 SD) | Severe DD and ID, sit with supported, no hand use, few words | Impaired sleep pattern | Yes | Axial hypotonia with limb spasticity, stereotypic movements, dyskinesia, dystonia, choreoathetosis, | Strabismus, eye contact and smiling intentionally |
| *Bay01/F* | 7 y 5 m | p.Gln86Argfs\*106 | <1 week / MIC, DD | ND | 47,5 cm (-5 SD) | Severe DD and ID, poor speech | Impaired sleep pattern | Absent | Axial hypotonia, hyperkinetic, stereotypical movements, orofacial dyskinesia, scoliosis | Strabismus, eye contact and smiling intentionally  |
| *Mon01/M* | 10 y 8 m | p.Gln86Argfs\*106 | 6 m/ Crying, weeping, intermittent esotropia, DD | 33 cm (-1,3 SD) | 46 cm (- 6 SD) | Severe DD and ID, able to sit unaided, no speech  |  Inappropriate laughing | Yes | Axial hypotonia, orofacial dyskinesia, limb dyskinesia, hand to mouth stereotypies  | Permanent esotropia, intermittent eye contact |
| *Str01/F* | 22 m | p.Pro105Argfs\*87 | 4 m/ DD  | 34 cm (-0,7 SD) |  -3,5 SD | Severe DD and ID, no acquisition, no hand use | Absent | Yes | Axial hypotonia with limb spasticity, stereotypical movements, dyskinesia, dystonia, choreoathetosis | Esotropia, eye fixation |
| *Rou01/M* | 3 y | p.Glu136Glyfs\*39 | 2 m/ Hypotonia, MIC | 34 cm (-0,7 SD) | 43,5 cm (-5,3 SD) | Severe DD and ID, no speech | Impaired sleep pattern, inappropriate laughing | Absent | Axia hypotonia, choreoathetosis | No strabismus, inconsistent eye contact  |
| *Ren01/M* | 12 y | p.Glu154Glyfs\*301 | 1 m/ MIC, development delay, esotropia | 31cm (-2.4 SD) | 48.5 cm (-4 SD) | Severe DD and ID, unsupported sitting posture (12 m), involuntary gross manipulation | Absent | Absent | Axial hypotonia with limb spasticity, hyperkinetic movement desorder, stereotypical movements (hand washing) | Permanent esotropia |
| *Leu01/F* | 5 y | p.Glu154Glyfs\*301 | 3 m/ Hypotonia | 31.7 cm (-1,7 SD) | 45 cm (-4,1 SD) | Severe DD and ID, friendly, smiling, unsupported sitting posture and step with help | Inappropriate laughing | Yes, feeding with solids | Axial hypotonia with limb spasticity, mild hyperkinesia, hand stereotypies | Eye contact, attention seeking, permanent esotropia |
| *Thi01/F* | 9 y | p.Glu154Glyfs\*301 | ND/ MIC DD | 34 cm (-0,7 SD) | 44,5cm (-5,8 SD) | Profound DD and ID, no speech | Absent  | Yes, mild  | Axial hypotonia with limb spasticity, Hyperkinetic movements, dystonia, stereotypic movements, orofacial dyskinesia, scoliosis | Permanent esotropia |
| *Im06/F* | 4 y 10 m | p.Glu154Glyfs\*301 | 3 m/ Hypotonia, MIC | 34 cm (-1,1 SD) | 43,5 cm (-5,2 SD) | Severe DD and ID, friendly, smiling, unsupported sitting posture, able to hold and manipulate | Impaired sleep pattern  | Yes, mild  | Axial hypotonia, orofacial dyskinesia, hyperkinesia, stereotypical movements (mild) | Eye contact, Permanent esotropia |
| *Im09/M* | 6 y | p.Glu154Glyfs\*301 | 7 m / Movement disorders, MIC | 33 cm (-1,3 SD) | 44 cm (-4,1 SD) | Severe DD and ID, and walk with help, able to hold and manipulate | Impaired sleep pattern  | Yes, mild  | Axial hypotonia with limb spasticity, limb dyskinesia, stereotypical movements | Attention seeking, permanent esotropia |
| *Ang02/M* | 2 y | p.Glu154Glyfs\*301 | 3 m/ Epileptic seizures  | 35 cm (-0,5 SD) | 44 cm (-3,5 SD) | Severe DD and ID, no acquisition | Impaired sleep pattern, inappropriate laughing and crying, screaming spells | Yes, difficulties with solids | Hypotonia, generalized dyskinesia with opisthotonos  | Cortical blindness, esotropia |
| *Im04/F* | 11 y | p.Glu155Glyfs\*300 | 2 m/ Esotropia, MIC, hypotonia, DD | 34 cm (-0,4 SD) | 45.5 cm (-3,2 SD) | Severe DD and ID, smiling intentionally | Inappropriate laughing and crying, screaming spells | Yes, percutaneous gastrostomy | Axial hypotonia with limb spasticity, erratic myoclonus, stereotypies of the median line, scoliosis | Esotropia, inconsistent eye contact |
| *Tou01/M* | ND | p.Glu155\* | 3 m/ Hypotonia | 33,5 cm (-1 SD) | 42,8 cm (-5,9 SD) | Severe DD and ID, no speech | Absent | Absent | Axial hypotonia, stereotypical movements | Strasbismus, intermittent eye contact  |
| *Im10/M* | 4 y 12 m | p.Lys162Serfs\*51 | 4 m/ DD,irritabiliy, absent visual contact, esotropia | 34 cm (-0,4 SD) | 49 cm (-1,8 SD) | Severe DD and ID, no speech | Inappropriate laughing | Yes  | Axial hypotonia with unstable head holding, severe generalized dyskinesia, dystonia, hand to mouth stereotypical movements | Permanent esotropia, amblyopia |
| *Rdb02/M* | 6 y | p.Tyr179\* | 5 m/ Truncal hypotonia | 34 cm (0 SD) | 45 cm (-5 SD) | Severe DD and ID | Severe impaired sleep pattern, inappropriate cryng | Absent | Axial hypotonia with limb spasticity, dyskinetic movement disorder, minor dysmorphic features | Strabismus, eye contact and smiling intentionally |
| *Im03/F* | 2 y 4 m | p.Ser185Glnfs\*270 | 5 m/ Hypotonia, DD | 32,5 cm (-1,2 SD) | 43 cm (-3,8 SD) | Severe DD and ID | Impaired sleep pattern | Yes  | Axial hypotonia with limb spasticity, dystonic movements, stereotypical movements | Esotropia, inconsistent eye contact smiling intentionally |
| *Pit02/M* | 22 y | p.Ile194Serfs\*19 | 3 m/ Hypotonia, poor eye contact, nystagmus | NE | NE | Severe DD and ID | Impaired sleep pattern, inappropriate laughing and crying, screaming spells | Yes, percutaneous gastrostomy | Axial hypotonia with limb spasticity, hyperkinetic movements | NE |
| *Ren03/M* | 32 y | p.Gln196\*  | 2 m/ DD, absence of ocular pursuit | 34 cm (-0,7 SD) | 50.5 cm (-4,5 SD) | Severe DD and ID, no speech | NE | NE | Axial hypotonia with limb spasticity, dystonia, choreoathetosis, scoliosis | NE |
| *Ade01/M* | 7 y | p.Tyr208\_Ile211del | Birth/MIC, DD | 25 cm (-1,9 SD) | 40.5 cm (-6 SD) | Severe DD and ID, no speech | Impaired sleep pattern | Yes, since birth, percutaneous gastrostomy | Axial hypotonia with limb spasticity, mixed hyperkinetic movements, dystonia, choreoathetosis, stereotypical movements | Eye contact, bilateral esotropia, smiling intentionally |
| *Str03/M* | 20 y | p.Val242Cysfs\*84 | 4 m/ DD, hypotonia | 33 cm (-0,6 SD) | 45 cm (-4,1 SD) | Severe DD and ID, sit with supported, no speech | Inappropriate laughing and crying, screaming spells | Absent | Dyskinesia, dystonia, choreoathetosis, stereotypical movements | Strabismus, eye contact and smiling intentionally |
| *Im01/F* | 10 y | p.Tyr254Thrfs\*72 | Birth/ MIC, hypotonia | 30 cm (-1,3 SD) | 45 cm (-2,6 SD) | Severe DD and ID | Impaired sleep pattern, inappropriate laughing and crying, screaming spells | Yes, since birth | Axial hypotonia with minor limb spasticity, orofacial dyskinesia, limb dystonia, stereotypical movement | Eye contact, permanent esotropia, and smiling intentionally |
| *Tel01/M* | 3 y 6 m | p.Ile266Tyrfs\*189 | 6 m/ MIC, DD | 33,5 cm (-0,7 SD) | 44 cm (-4,5 SD) | Severe DD and ID, sit with supported, few babbling | Absent | Absent | Moderate axial hypotonia, hyperkinetic movements | Strabismus |
| *Ren02/F* | 10 y | p.Pro182Leu | 1 m/ Opisthotonus, DD, esotropia | 30 cm (-2,5 SD) | NE | Moderate DD and severe ID, sit with supports, loss of hand use, some babbling | Inappropriate laughing | Yes | Axial hypotonia with feet spasticity, dyskinesia, dystonia, choreoathetosis | Strabismus, eye contact and smiling intentionally |
| *Rdb01/M* | 8 y | p.Asn187Asp | 3 m/ Seizures | 31 cm (-2,1 SD) | 46 cm (-3,3 SD) | Severe DD and ID, sit with supported, no speech | Impaired sleep pattern | Absent | Axial hypotonia with limb spasticity, stereotypical movements, dyskinesia, dystonia, choreoathetosis | Strabismus, eye contact and smiling intentionally |
| *Lau01/M* | 2 y | p.Asn187Lys | 12 days/ FS with apneas | 34 cm (-0,4 SD) | 50.5 cm (-1,1 SD) | Severe DD and ID, no speech | Impaired sleep pattern | Yes, percutaneous gastrostomy | Axial hypotonia, stereotypical movements (orofacial and head), orofacial dyskinesia, choreoathetosis | No strabismus, inconsistent eye contact, and smiling intentionally  |
| *Im08/M* | 22 m | p.Arg195Pro | Prenatal diagnosis of MIC with thick corpus callosum | 33,5 cm (-1,7 SD) | 45 cm (-4,1 SD) | Severe DD and ID, sit with supported, able to hold and manipulate objects  | Impaired sleep pattern, Intermittent inappropriate laughing and crying and screaming spells | Yes  | Axial hypotonia with limb spasticity, hyperkinesia | No strabismus, intermittent eye contact, and smiling intentionally  |
| *Pit01/F* | 42 y | p.Leu204Phe | 3 m/ MIC, DD, feeding difficulties |  - 1 SD | 50.5 cm (-3,5 SD) | Severe DD and ID, sit with supported and walk independently, no speech acquisition  | Inappropriate laughing and crying, screaming spells | Absent | Axial hypotonia with limb spasticity, hyperkinetic movements, dystonia, choreoathetosis, stereotypical movements, orofacial dyskinesia | Permanent esotropia, amblyopia |
|  *Mar01/F* | 4 y | p.Phe215Leu | 2 m/ Hypotonia, esotropia | 34 cm (-0,7 SD) | 46 cm (-4,4 SD) | Severe DD and ID, sit with supported, walk with help, complete loss of acquired hand use, few babbling  | Absent | Yes  |  Limb spasticity, hyperkinetic dyskinetic movements, orofacial dyskinesia, stereotypical movements | Strabismus |
| *Im12/F* | 2 y 7 m | p.Gly224Ser | 3 m/ Erratic ocular pursuit, hypotonia | 33.5 cm (-0,7 SD) | 47 cm (-1,2SD) | Moderate DD, severe ID, sit with supported | Impaired sleep pattern, inappropriate laughing and crying, screaming spells | Yes, minor | Hypotonia, stereotypical hand movements | Eye contact and smiling very limited at 3-4 months, then lost until after 12 months; smile with sensory and auditory stimuli;  |
| *Ang01/M* | 2 y 4 m | p.Arg230His | 4 m/ DD, axial dystonia | 31,5 cm (-2,5 SD) | 42 cm (-6,2 SD) | Severe DD and ID, no speech | Impaired sleep pattern, inappropriate laughing and crying, screaming spells | Yes  | Axial hypotonia, stereotypical movements, dyskinesia, choreoathetosis, dystonia | Strabismus, eye contact and smiling intentionally |
| *Str02/F* | 9 y | p.Gly252Val | 6 m/ Esotropia | 36 cm (0,1 SD) | 47,5 cm (-3,8 SD) | Severe DD and ID, sit with supported, few words |  Inappropriate laughing and crying, screaming spells | Absent | Axial hypotonia with limb spasticity, stereotypical movements, movement disorders | Strabismus, eye contact and smiling intentionally |
| *Lyo01/M* | 4 y | p.Trp255Arg | 3 m/ MIC, esotropia, growth delay, DD | 35 cm (-0,5 SD) | NE | Severe DD and ID, no speech | Rare inappropriate laughing and crying, and, screaming spells | Yes | Hypotonia, hand to mouth stereotypies, permanent dystonic movements, scoliosis  | Strabismus, eye contact and smiling intentionally |
| *Nan03/F* | 17y |  p.Leu257Pro | 4 m/ hypotonia, no visual fixation | 34 cm (-0,5 SD) | 50,3 cm (-3 SD) | Severe DD and ID, no speech | Absent | Initial feeding difficulties, but no gastrostomy required | Axial hypotonia with limb spasticity, stereotypical movements on the median line, dystonic movements, scoliosis, strabismus | Strasbismus, intermittent eye contact  |
| *Nan01/F* | 7 y 1 m | p.Asn408Ile | 14 m/ DD | 34 cm (0 SD) | 48,5 cm (-2,2 SD) | Severe DD and ID, sit without supported, walk independently, few words | Impaired sleep pattern, inappropriate laughing and crying, screaming spells | Yes, since birth | Axial hypotonia, stereotypical movements, dyskinesia, choreoathetosis, dystonia | Strabismus, eye contact and smiling intentionally |
| *Im02/M* | 4 y 9 m | p.Ser326Glufs\*129 | 6 m/ MIC, movement disorders | 35,5 cm (0,5 SD) | 45 cm (-5,2 SD) | Severe DD and ID, few babbling | Impaired sleep pattern, inappropriate laughing and crying, screaming spells | Yes  | Axial hypotonia, severe limb and orofacial dyskinesia, hand to mouth stereotypies | Esotropia, inconsistent eye contact, smiling intentionally |
| *Cle01/F* | 18 y | p.Tyr400\* | 6 m / DD | 34 cm (-1,1 SD) | 50,5 cm (-3,5 SD) | Severe DD and ID, sit without supported, walk independently, no speech | Impaired sleep pattern, inappropriate laughing and crying, screaming spells | Absent | Axia hypotonia, choreoathetosis, dyskinesia, dystonia, stereotypical movements, scoliosis | Strabismus, eye contact and smiling intentionally |
| *Dij01/F* | 18 y 2 m | c.-715delinsTACCAAAA​ | 6 m/ MIC, DD, hypotonia, poor eye contact | 33 cm (-1,7 SD) | 50 cm (-3 SD) | Severe DD and ID, no speech | Impaired sleep pattern | Yes | Axial hypotonia with limb spasticity, pyramidal syndrome, stereotypical movements  | Yes |
| *Lyo02/F* | 1 y 6 m | 14q12(29,222,002-29,258,618) | Birth / Nystagmus | 32,4 cm (-1,3 SD) | 43 cm (-4,8 SD) | Severe DD and ID, no speech | Impaired sleep pattern | Yes | Axial hypotonia with limb spasticity, intermittent permanent limb dyskinesia | Strabismus, eye contact and smiling intentionally |
| *Lyo03/F* | 3 y 9 m | 14q12(26,415,516-29,677,148) | 3 m/ Hypotonia, MIC | 31,1 cm (-2 SD) | 42 cm (-5,3 SD) | Severe DD and ID, no speech |  Inappropriate laughing and crying | Yes, percutaneous gastrostomy | Axial hypotonia, intermittent proximal limb choreoathetosis, scoliosis | Strabismus, eye contact and smiling intentionally |
| *Im07/F* | 2 y | del14q12q13.1 | Prenatal diagnosis of corpus callosum agenesis | 32 cm (-1,9 SD) | 41 cm (-5,1 SD) | Severe DD and ID, no speech | Severe impaired sleep pattern | Yes, since birth, percutaneous gastrostomy | Axial hypotonia, orolingual dyskinesia, hyperkinesia, hand to mouth stereotypical movements | Esotropia and intermittent eye contact and gaze |
| *Aix01/M* | 7 y | del14q(18,798,641\_19,484,013) | Prenatal diagnosis of corpus callosum agenesis | 31,5 cm (-0,8 SD) | 45 cm (-5,3 SD) | Severe DD and ID, no speech | Severe impaired sleep pattern, inappropriate laughing  | Yes, since birth, with severe gastrooesophageal reflux  | Axial hypotonia with no head control and limb spasticity, severe generalized dyskinesia, stereotypical movements, dysmorphic features | Esotropia |

Abbreviations: DD = developmental delay; F = female; ID = intellectual disability; m = months; M = male; NE = not evaluated; OFC = occipitofrontal head circumference; MIC: microcephaly (defined as < 2 SD score according to country-specific control cohorts; WHO child growth standards and the Fenton growth chart for preterm infants).