Table S3 Differential Diagnosis

	Gene / Locus	Inheritance	Speech and language development	Neurologic development
FOXP1-related ID syndrome	FOXP1	Autosomal dominant	Severe delayed speech development - Expressive language disorder - OMD - Articulation problems	ID - NMD
Speech-language disorder 1 (SPCH1)	FOXP2	Autosomal dominant	DVD - OMD - Impaired processing and expression of language - Articulation defects	Decreased nonverbal IQ
Specific language impairment 1 (SLI1)	-	Multifactorial	SLI - May affect language expression, reception, and/or articulation	Normal intelligence - No other neurologic deficits
Specific language impairment 2 (SLI2)	-	Multifactorial	SLI - May affect language expression, reception, and/or articulation	Normal intelligence - No other neurologic deficits
Specific language impairment 3 (SLI3)	-	-	SLI	-
Specific language impairment 4 (SLI4)	-	-	SLI	-
Specific language impairment 5 (SLI5)	TM4SF20	Autosomal dominant	SLI - Early speech delay - Communication defects	GDD might be present - Others show normal cognitive function
Familial developmental dysphasia	-	Autosomal dominant	Developmental dysphasia - Specific and severe delay in development of spoken language	Normal intelligence
Distal chromosome 11p13 deletion syndrome	-	Autosomal dominant	Speech and language delay	Developmental delay - ID
Phelan-Mcdermid syndrome (PHMDS)	SHANK3	Isolated cases	Absent or delayed speech development - Severe compromised expressive language development	GDD - Moderate to severe ID - Generalized hypotonia
Floating-harbor syndrome (FLHS)	SRCAP	Autosomal dominant	Expressive language delay	Mild ID versus normal intelligence - Normal motor development
Helsmoortel-van der aa syndrome (HVDAS)	ADNP	Autosomal dominant	Language impairment	Developmental delay - Delayed psychomotor development - Hypotonia - Mild to severe ID
Bainbridge-ropers syndrome (BRPS)	ASXL3	-	Language impairment	Severe psychomotor retardation

	Behavioral trouble	Craniofacial features	Associated organ involvement
FOXP1-related ID syndrome	Autistic behavior - ASD - Aggression - Obsessions Compulsions - Hyperactivity	Typical dysmorphic features	CHD - Feeding difficulties - GTA - Ophtalmological problems
Speech-language disorder 1 (SPCH1)	-	-	-
Specific language impairment 1 (SLI1)	-	-	-
Specific language impairment 2 (SLI2)	-	-	-
Specific language impairment 3 (SLI3)	-	-	-
Specific language impairment 4 (SLI4)	-	-	-
Specific language impairment 5 (SLI5)	ASD	Normal facial appearence	No associated congenital defects
Familial developmental dysphasia	-	-	-
Distal chromosome 11p13 deletion syndrome	Autism	-	Congenital eye malformations - Aniridia
Phelan-Mcdermid syndrome (PHMDS)	Autistic features - Poor social interaction - Aggressive behavior - Inappropriate chewing behavior	Normal to accelerated growth - Tall stature - Macrocephaly - Typical dysmorphic features versus normal facial appearence	Feeding difficulties
Floating-harbor syndrome (FLHS)	-	Short stature - Typical dysmorphic features (age-related)	CHD (rare) - Umbilical hernia (rare) - Celiac disease - GTA - Renal abnormalities - Skeletal abnormalities
Helsmoortel-van der aa syndrome (HVDAS)	ASD - Obsessive-compulsive behavior - Stereotypic behavior - Hyperactivity	Short stature - Obesity - Typical dysmorphic features	CHD - Feeding difficulties - Joint laxity
Bainbridge-ropers syndrome (BRPS)	Stereotypic behavior - Autistic features	Severe postnatal growth retardation - Dysmorphic features	Feeding problems - Ulnar deviation of hands