

List of Figures

1.1	Conceptual framework of human functioning	3
1.2	Etiology of Intellectual Disability	7
1.3	Sequence organization of telomeres	23
1.4	Current practice of evaluating a subject with ID	27
1.5	Representative images of a few dysmorphic features	30
2.1	Karyotype of normal female	66
2.2	Karyotype of normal male	67
2.3	FXS screening: analysis of female samples	70
2.4	A composite FISH karyogram	73
2.5	MLPA: normal result	76
2.6	MLPA: abnormal peak ratio	77
2.7	QMPSF: normal result	79
2.8	qPCR amplification plot showing amplification in all sample wells	86
2.9	Standard curve construction using Ct values vs. log of known DNA concentrations	86
3.1	Karyotype:46,XX,inv(17)	93
3.2	Karyotype: 46,XX,del(18p)	93
3.3	Karyotype: 46,XY,del(18q)	95
3.4	Karyotype: 46,XY,del(18q)	96
3.5	FXS screening: PCR1 results	97
3.6	FXS screening: PCR2 results	98
4.1	Subtelomeric FISH: del(18pter)	108
4.2	Subtelomeric FISH: del(18qter)	109
4.3	Subtelomeric FISH: del(18qter)	110
4.4	Subtelomeric FISH: der(7)	111
4.5	Subtelomeric FISH: t(3qter;7qter)	112
4.6	Subtelomeric FISH: del(17pter)	113
4.7	Subtelomeric FISH: der(Y)	114
4.8	Subtelomeric FISH: der(1)	115
4.9	Subtelomeric FISH: del(4pter)	116
4.10	Subtelomeric FISH: der(4)	118
4.11	Subtelomeric FISH: t(4pter;8pter)	119
4.12	Subtelomeric FISH: der(6)	120

5.1	MLPA:15q11 deletion	130
5.2	MLPA: 7q11.23 deletion	131
6.1	MLPA: duplication 3qter and deletion 7qter	141
6.2	MLPA: deletion Yqter	142
6.3	qPCR: Gene dosage ratios	146