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### LIST OF ABBREVIATIONS AND SYMBOLS

add = additional material of unknown origin

AS = Angleman syndrome

BAC = Bacterial Artificial Chromosome

bp = base pair(s)

iC = degree celsius

CARS = The Childhood Autism Rating Scale

CCD = charged coupled device

CGH = Comparative Genomic Hybridization

DAPI = 4,6-Diamino-2-Phenyl-indole

dATP = deoxyadenosine-5'-triphosphate

dCTP = deoxycytidine-5'-triphosphate

dGTP = deoxyguanosine-5'-triphosphate

dTTP = deoxythymidine-5'-triphosphate

dUTP = deoxyurasine-5'-triphosphate

dNTP = dATP + dCTP + dGTP + dTTP

del = deletion

der = derivative chromosome

DF = dysmorphic features

DMSO = dimethylsulfoxide

DNA = deoxyribonucleic acid

dn = de novo

dup = duplication

et al. = et ali (Latin) and others

FH = family history of mental retardation

FISH = Fluorescence *in situ* Hybridization

FMR1 = Fragile X mental retardation 1

FMR2 =  $Fragile\ X\ mental\ retardation\ 2$ 

FXS = Fragile X syndrome

GTG = Giemsa-Banding Technique Using Trypsin

ICD-10 = The International Classification of Diseases,

10<sup>th</sup> Edition

idic = isodicentric chromosome

## LIST OF ABBREVIATIONS AND SYMBOLS (Continued)

IMR = idiopathic mental retardation

inv = inversion

IQ = intelligence quotient

ISCN = International Standing Committee on Human

Cytogenetic Nomenclature

ish = in situ hybridization

Kb = kilobase(s)

M = molar

mar = marker chromosome

Mb = mega base pair(s)

mM = millimolar

MCA = multiple congenital anomalies

ml = milliliter

mm = millimeter

mos = mosaic

MAPH = multiplex amplifiable probe hybridization

MLPA = multiplex ligation dependent probe amplification

MR = mental retardation

M-TEL = multiplex FISH telomere integrity assay

MTX = methotrexate

p-arm = short arm of chromosome

PAC = P1-derived Artificial Chromosome

PCR = polymerase chain reaction

PDD = Pervasive Developmental Disorders

PDD-NOS = Pervasive Developmental Disorder Not Otherwise

Specific

PHA = phytohemagglutin

PKU = phenylketonuria

PWS = Prader-Willi syndrome

q-arm = long arm of chromosome

RNA = ribonucleic acid

r = ring chromosome

# LIST OF ABBREVIATIONS AND SYMBOLS (Continued)

RELN = Reelin

SDS = sodium dodecyl sulfate

SKY = spectral karyotype

SSC = standard saline citrate

t = translocation

TAR = telomere associated repeat region

TSC = tuberous sclerosis complex

VCF = Velocardio-Facial Syndorme

 $\mu l$  = microliter

 $\mu$ M = micromolar

U = unit(s)

UV = ultraviolet