Case ID: 4161

Personal data:

Clinician ID: 338

Clinician Name: Bon, B van

Clinician Centre: THE NETHERLANDS: Radboud University Nijmegen Medical Centre, Department of Human Genetics

Cytogeneticist ID: 10

Cytogeneticist Name: Leeuw, Nicole de

Cytogeneticist Centre: THE NETHERLANDS: Radboud University Nijmegen Medical Centre, Department of Human Genetics

Date of birth: 2000-02-28

Sex: Female

Original country: The Netherlands

Case characteristic:

The case is from ECARUCA Submission.

Is the case certain? true

Last update date: 2013-02-28

Case status:

2005-12-29	New Case
2006-01-02	Pre-Registered
2007-08-17	Registered
2007-09-28	Accepted
2007-10-05	Completed

Is follow up possible?: true

Clinical data:

Age at last exam: 5 year 9 month.

Level of mental retardation estimated at age:

60 months Severe (IQ < 30)

Clinical feature:

Stature:

Short stature,general

Cranium:

Microcephaly

Ears:

Ear helix,general abnormalities

Eyes,associated structures:

Blepharophimosis

Epicanthic folds

Nose:

Depressed/flat nasal bridge

Wide nasal bridge

Face:

Flat face

Mouth:

Down-turned corners of the mouth

Long philtrum

Thin upper lip

Oral region:

Cleft uvula

Back and spine:

Scoliosis

Hands:

Camptodactyly

Neurology:

Hypotonia

Abdomen:

Gastro-oesophageal reflux

Hands:

Short 5th finger

Other clinical investigations:

Test date:	2001-12-01
Test name:	EEG
Test Result:	no abnormalities
Test date:	2000-10-09
Test name:	Metabolic Testing
Test Result:	organic acids no abnormalities
	aminoacids no abnormalities
	variable lactate values (normal-high)
Test date:	2000-10-09
Test name:	Xray leg
Test Result:	tibia femur index: 0,75 disproportional
Test date:	2000-10-08
Test name:	Xray spine/hips/thorax
Test Result:	no abnormalities

Test date: 2000-10-01

Test name: Cerebral MRI Scan

Test Result: no abnormalities

Test date: 2000-10-01

Test name: echo cerebrum

Test Result: enlarged ventricles

Cytogenetic data:

Mother karyotype:

unknown

Father karyotype:

46,XY,t(10;12)(q26;p13.3)

ISCN result:

46,XX,der(12)t(10;12)(q25.3;p13.31)pat.arr 10q25.3q26.3(118,567,107-135,427,144)x3,12p13.33p13.31(173,787-5,685,642)x1 (hg19)

Additional cytogenetic remark:

Affymetrix CytoScan HD array platform (Humane Genome Build hg19/2009)

Result of paternal balanced translocation. Another 3 patients in

family with unbalanced der(12)

Aberrations:

Aberration type duplication (dup)

Chromosome number 10

Breakpoint1 q25.3

Breakpoint2 q26.3

Breakpoint accuracy:

Remark1: translocation

Remark2:

Remark3: microarray

Remark4:

Remark5:

Aberration type deletion (del)

Chromosome number 12

Breakpoint1 p13.33

Breakpoint2 p13.31

Breakpoint accuracy:

Remark1: translocation

Remark2:

Remark3: microarray

Remark4:

Remark5:

~~~~~~

Material: Blood Lymphocytes

Routine chromosome analysis: Routine GTG-banding

Karyotype: 46,XX,der(12)t(10;12)(q26;p13.3)pat

Molecular data:

~~~~~~