Case ID: 4818 ***RING CHR, MOSAICISM***

Personal data:

Clinician ID: 691

Clinician Name: Ravenswaaij, Conny van

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

Cytogeneticist ID: 1148

Cytogeneticist Name: Leegte, Beike

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

Date of birth: 2004-01-18

Sex: Female

Original country: The Netherlands

Case characteristic:

The case is from ECARUCA Submission.

Is the case certain? true

Last update date: 2012-05-15

Case status:

2011-03-24	New Case
2011-06-20	Pre-Registered
2012-05-03	Registered
2012-05-07	Accepted
2012-05-15	Completed

Is follow up possible?: true

Clinical data:

Age at last exam: 7 year 10 month.

Level of mental retardation estimated at age:

94 months Moderate (IQ 30 - 50)

65 months Moderate (IQ 30 - 50)

Level of motor milestone estimated at age:

36 months Walk

22 months Sit Independently

22 months First Words

Clinical feature:

Cranium:

Brachycephaly

Eyes,associated structures:

Palpebral fissures slant up

Mouth:

Down-turned corners of the mouth

Oral region:

Cleft uvula

Hands:

Clinodactyly

Neurology:

Abnormal gait

Hypotonia

Mental retardation

Seizures/Abnormal EEG

Gestation/delivery.:

Gestation,37-42 weeks.

Other clinical investigations:

Test date: 2004-09-01

Test name: EEG

Test Result: left temporal epileptic focus

Test date: 2004-07-04

Test name: opthalmology

Test Result: normal

Test date: 2004-07-02

Test name: Cerebral MRI Scan

Test Result: delayed myelinisation frontal

Test date: 2004-07-01

Test name: Metabolic Testing

Test Result: normal

Test date: 2004-06-01

Test name: EEG

Test Result: West syndrome

Cytogenetic data:

Mother karyotype:

46,XX

Father karyotype:

46,XY

ISCN result:

47,XX,+r(14)(::q11.2->q21.3::)dn[9]/46,XX[21].ish r(14)(wcp14+,D14Z1/D22Z1-,P820M16-)(RP11-68m15+,RP11-1083E2).arr 14q11.2q21.2(22,235,782-44,973,797)x2~3 (hg19)

Additional cytogenetic remark:

47,XX,+r(14)(::q11.2->q21.3::)dn[9]/46,XX[21].ish

r(14)(wcp14+,D14Z1/D22Z1-,P820M16-)(RP11-68m15+,RP11-1083E2).arr cgh

14q11.2q21.2(rp11-92f11->rp11-99l13)x2~3 (hg18)

Aberrations:

Aberration type duplication (dup)

Chromosome number 14

Breakpoint1 q11.2

Breakpoint2 q21.2

Breakpoint accuracy:

Remark1: ring , mosaic

Remark2:

Remark3: microarray

Remark4:

Remark5:

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Material: Blood Lymphocytes

Routine chromosome analysis: GPG-banding

ISCN quality: 550

Karyotype: mos

47,XX,+r(14)(::q11.2->q21.3::)[9]/46,XX[21]

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Material: Blood Lymphocytes

Routine chromosome analysis: FISH

ISCN quality: 350

Karyotype: ish r(14)(wcp14+,D14Z1/D22Z1-,P820M16-) ish r(14)(RP11-68m15+,RP11-1083E2)

Molecular data:
