Case ID: 4160

Personal data:

Clinician ID: 9

Clinician Name: Ravenswaaij, Conny van

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

Cytogeneticist ID: 11

Cytogeneticist Name: Ravenswaaij, Conny van

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

Date of birth: 2001-03-27

Sex: Female

Original country: The Netherlands

Case characteristic:

The case is from ECARUCA Submission.

Is the case certain? true

Last update date: 2013-02-25

Case status:

2005-12-29	New Case
2006-01-02	Pre-Registered
2006-02-27	Registered
2006-03-06	Accepted

2006-03-06 Completed

Is follow up possible?: true

Clinical data:

Age at last exam: 4 year 9 month.

Level of mental retardation estimated at age:

57 months Severe (IQ < 30)

30 months Moderate (IQ 30 - 50)

Level of motor milestone estimated at age:

24 months Walk

21 months Sit Independently

Clinical feature:

Stature:

Short stature,proportionate

Eyes,globes:

Hypertelorism

Eyes,associated structures:

Ptosis of eyelids

Short palpebral fissures

Nose:

Upturned nose

Mouth:

Down-turned corners of the mouth

Oral region:

Cleft palate

Back and spine:

Sacral dimple/sinus

Thorax:

Ventricular septal defect

Feet:

High arches of feet

Haematol/Immunology:

Recurrent infections

Neurology:

Mental retardation

Other clinical investigations:

Test date: 2003-08-21

Test name: Cerebral MRI Scan

Test Result: normal

Cytogenetic data:

ISCN result:

46,XX,der(4)t(4;16)(p16.3;p12.3)[6]/46,XX[24].arr 16p13.3p12.3(60,001-17,185,313)x2~3 (hg19)

Additional cytogenetic remark:

46,XX,der(4)t(4;16)(p16.3;p12.3)[6]/46,XX[24].arr 16p13.3p12.3(1-17092814)x2~3 (hg18)

FISH: no deletion of WHS-region on 4p16.3

Microarray: Agilent 180 K custom HD-DGH; AMADID-23363 (hg18).

We adjusted the karyotype in accordance with ISCN 2009 (hg19)

Aberrations:

Aberration type duplication (dup)

Chromosome number 16

Breakpoint1 p13.3

Breakpoint2	p12.3
Breakpoint accuracy:	
Remark1:	mosaic 20%
Remark2:	
Remark3:	microarray
Remark4:	
Remark5:	

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Material:	Blood Lymphocytes
Routine chromosome analysis:	Routine GTG-banding
Karyotype:	46,XX,der(4)(t(4;16)(p16.3;p13.1)/46,XX
	[6/24]

Other molecular cytogenetic methods:

MLPA

Molecular data:

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