Case ID: 4988

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: 2011-01-23

Sex: Male

Original country: The Netherlands

Case characteristic:

The case is from ECARUCA Submission.

Is the case certain? true

Last update date: 2013-10-25

Case status:

2013-01-28	New Case
2013-01-28	Pre-Registered
2013-08-19	Registered
2013-09-20	Accepted
2013-10-25	Completed

Is follow up possible?: false

Follow up impossible reason:

Prenatal case; terminated pregnancy

Clinical data:

Clinical feature:

Stature:

Short stature,prenatal onset

Eyes,globes:

Hypertelorism

Prominent eyes (inc. proptosis/shallow orbits)

Face:

Cleft mandible

Small mandible/micrognathia

Mouth:

Cleft upper lip (non-midline)

Back and spine:

Scoliosis

Thorax:

Congenital hernia of diaphragm

Ventricular septal defect

Lung hypoplasia/agenesis

Pulmonary segmentation defects

Abdomen:

Polysplenia

Genitalia:

Hypospadias

Feet:

Club foot, varus

Other clinical investigations:

Test date: 2011-01-23

Test name: obduction

Test Result: no brain abnormalities

Pedigree data:

Mother balanced translocation chromosome 3 and 4, father of mother

also balanced translocation

Cytogenetic data:

Mother karyotype:

46,XX,t(3;4)(p25.1;p15.33)

Father karyotype:

46,XY

ISCN result:

46,XY,der(4)t(3;4)(p25.1;p15.33)mat.arr 3p26.3p25.1(61,875-13,407,536)x3,4p16.3p15.33(106,688-13,767,891)x1 (hg19)

Additional cytogenetic remark:

46,XY,der(4)t(3;4)(p25.1;p15.33)mat.arr 3p26.3p25.1(36,875-13,382,536)x3,4p16.3p15.33(96,688-13,376,989)x1 (hg18)

13.3 Mb gain in 3p26.3p25.1 and 13.3Mb loss in 4p16.3p15.33

Prenatal case; terminated pregnancy

Aberrations:

Aberration type duplication (dup)

Chromosome number	3
Breakpoint1	p26.3
Breakpoint2	p25.1
Breakpoint accuracy:	
Remark1:	
Remark2:	
Remark3:	microarray
Remark4:	
Remark5:	

Aberration type	deletion (del)
Chromosome number	4
Breakpoint1	p16.3
Breakpoint2	p15.33
Breakpoint accuracy:	
Remark1:	
Remark2:	
Remark3:	microarray
Remark4:	
Remark5:	

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Material:	Chorion Villi
Culture conditions:	
Standard Media	
Media Supplements	Not applicable
Routine chromosome analysis:	Routine GTG-banding
Number of cells examined:	9999
ISCN quality:	350
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Karyotype:	46,XY (due to insufficient quality of the chromosomes, the unbalanced translocation was not detected at first analysis)

Molecular data:
