

CASE REPORT

Prenatal Phenotype Analysis of a Case with Cardiofaciocutaneous Syndrome Caused by BRAF Gene Mutation

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ABSTRACT

Background: This study analyzed the prenatal clinical phenotype of a child with cardiocutaneous syndrome (CFCS) caused by BRAF gene mutation.

Methods: Chromosome microarray (CMA) and whole exome sequencing (WES) were used for the prenatal genetic diagnosis in a pregnant woman with fetal bone dysplasia (fetal macrocephaly, short femur) and polyhydramnios.

Results: CMA revealed no abnormalities, WES revealed BRAF de novo missense mutation (c.1406G > A, p.G469E), and the fetal parents showed no BRAF gene mutation.

Conclusions: Pathogenic variants in the BRAF gene leads to CFCS, which during pregnancy presents with signs of ultrasonographic abnormalities including macrocephaly, short femur, polyhydramnios.
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Supplementary Data

Table S1. Ultrasonography shows the values of biparietal diameter, head circumference, abdominal circumference, femur length and the maximum depth of amniotic fluid in different gestation weeks.

Gesta- tion weeks (W)	Bipari- etal diameter (cm)	Bipari- etal diameter (Per- centile)	Head circum- ference (cm)	Head circum- ference (Per- centile)	Abdo- minal circum- ference (cm)	Abdomi- nal circum- ference (Per- centile)	Femur length (cm)	Femur length (Per- centile)	Maximum depth of amniotic fluid (mm)	Status of amniotic fluid
19	42	50%	160	50%	142	≤ 90%	26	≥ 10%	60	normal range
23	64	≥ 90%	219	≥ 90%	201	≥ 90%	36	= 10%	97	polyrhy- dramnios
26	71	≥ 90%	252	≥ 90%	223	≥ 90%	44	= 10%	125	polyrhy- dramnios
28	74	Close to 90%	266	≥ 90%	238	≥ 90%	47	= 10%	95	polyrhy- dramnios
30	79	Close to 90%	274	≥ 90%	265	≥ 90%	52	= 10%	114	polyrhy- dramnios
32	90	≥ 90%	312	≥ 90%	299	≥ 90%	54	≤ 10%	92	polyrhy- dramnios
34	90	≥ 90%	315	≥ 90%	312	≥ 90%	58	≤ 10%	113	polyrhy- dramnios
36	98	≥ 90%	347	≥ 90%	350	≥ 90%	59	≤ 10%	130	polyrhy- dramnios
38	99	≥ 90%	366	≥ 90%	377	≥ 90%	64	≤ 10%	138	polyrhy- dramnios

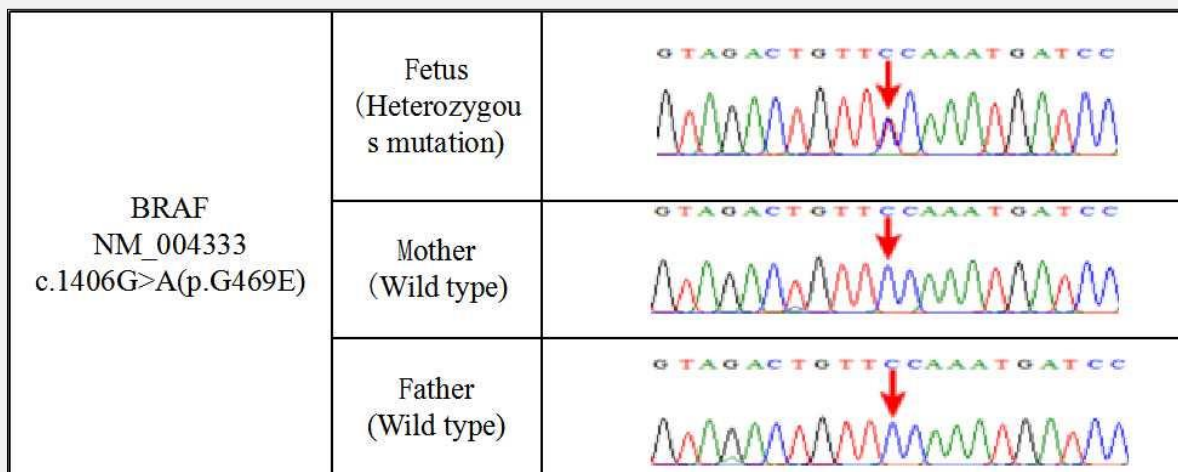


Figure S1. Sanger sequencing to verify the WES results, suggesting the presence of a de novo missense mutation, c. 1406G > A, in the fetal BRAF gene.