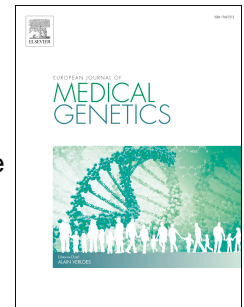


# Accepted Manuscript

Redefining the phenotypic spectrum of *de novo* heterozygous *CDK13* variants: Three patients without cardiac defects

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Table 1. Summary of the patients with developmental delay with or without congenital cardiac diseases and mutations in *CDK13*

Patient Age	Patient 1 7 years	Patient 2 3.5 years	Patient 3 5 years	16 patients reported by Sifrim et al. and Bostwick et al. 0.2 ~ 38 years	Summary
Congenital cardiac diseases	absent	absent	absent	13/16	13/19
Age reported/Sex	male	female	male	7 male and 9 female patients	9 male and 10 female patients
Gestational age	37 weeks	38 weeks	36 weeks	range: 35 ~ 41 weeks (among 5 available cases)	range: 35 ~ 41 weeks (among 8 available cases)
Birth weight	1705 g (-3.0 SD)	2650 g (-0.8 SD)	2380g (-1.5 SD)	range: 1700 ~ 3640 g (-2.7 ~ +1.2 SD) (among 5 available cases)	range: 1700 ~ 3640 g (-3.0 ~ +1.2 SD) (among 8 available cases)
Height/Length at diagnosis	103.2 cm (-2.7 SD)	87.8 cm (-0.9 SD)	112.5 cm (+0.2 SD)	range: (-3.8 ~ +0.2 SD) (among 5 available cases)	range: (-3.8 ~ +0.2 SD) (among 8 available cases)
Developmental delay/ Intellectual disability	present	present	present	16/16	19/19
Dysmorphic features	Eyes	hypertelorism, upslanting palpebral fissures	hypertelorism, upslanting palpebral fissures	hypertelorism (11/13), upslanting palpebral fissures (9/15)	hypertelorism (14/16), upslanting palpebral fissures (12/18)
	Nose	short and broad columella, wide nasal bridge and nasal ridge	short and broad columella, wide nasal bridge and nasal ridge	short and broad columella, wide nasal bridge and nasal ridge  (retrospective review of patients whose photos were available in previously reported documents)  short and broad columella (13/13), wide nasal bridge (11/13), wide nasal ridge (11/13)	broad and short columella (16/16), wide nasal bridge (14/16), wide nasal ridge (14/16)
	Ears	hypoplastic antihelix	hypoplastic antihelix, low-set ears, hearing impairment	posteriorly-rotated ears, low-set ears	low-set ears (8/15) dysplastic ears (11/11) hearing impairment (1/7)
	Teeth	peg-shaped, wide-spaced	absent	peg-shaped, wide-spaced	(not described previously)
Clinodactyly or Camptodactyly	clinodactyly of the 5 <sup>th</sup> finger	clinodactyly of the 5 <sup>th</sup> finger	clinodactyly of the 5 <sup>th</sup> finger	10/16	13/19

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