

CORRECTION OPEN



Correction: Clinical and genetic analysis further delineates the phenotypic spectrum of *ALDH1A3*-related anophthalmia and microphthalmia

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In this article, due to a typesetting mistake, the layout of Table 1 was wrong and it appeared incorrectly only in the article PDF; the table should have appeared as shown below.

Table 1. Summary of phenotypic and genetic findings.

Findings	Family 1, II:2	Family 1, II:4	Family 2, II:1	Family 3, II:1	Family 4, II:1	Family 5, II:1	Family 6, II:2	Family 6, II:4	Family 7, II:1
General information									
Age	25 y	15 y	27 y	9 y	29 y	3 y	9 y	Birth	18 m
Gender	M	M	M	F	M	M	F	F	M
Ethnicity	Asian	Asian	White British	Mixed European Origin	Caucasian	Moroccan	Libyan	Libyan	Spanish
Consanguinity	No	No	No	No	No	Yes	Yes	Yes	NK
Genetic findings									
ALDH1A3 variants ^a	c.874G>T, p.Asp292Tyr; c.1393A>T, p.Ile465Phe	c.874G>T, p.Asp292Tyr; c.1393A>T, p.Ile465Phe	c.845G>C, p.Gly282Ala; c.1459A>G, p.Arg487Gly	c.847_849del, p.Gly283del; c.953C>A, p.Ser318Tyr	c.566G>A, p.Trp189*; c.100- 2A>G	c.1233 + 2T>C	c.1144G>A, p.Gly382Arg	c.1144G>A, p.Gly382Arg	c.434C>T, p.Ala145Val
Growth									
Birth weight (kg)	3.45	2.38	2.77	3.1	3.2	3.6	3.3	3.77	NK
Height (cm, centile)	165.5, 2nd–9th (24 y)	156, 9th (14 y)	NK	48.5; 25th (birth)	172; 25th (25 y)	51; 50th (birth)	123; 9th (9 y)	50; 50th (birth)	NK
Weight (kg, centile)	76.5, 75th–91st (24 y)	35.6, 2nd (14 y)	NK	NK	NK	NK	24; 9th–25th (9 y)	NK	NK
HC (cm, centile)	58, 91st–98th (24 y)	52.3, 0.4th–2nd (14 y)	NK	32.5; 9th (birth)	59.2; 91st–98th (25 y)	37; 75th (birth)	50.3; 10th (9 y)	35.5; 91st (birth)	NK
Ocular									
Anophthalmia	Bil	Bil	Bil	-	-	-	Bil	Bil	-
Microphthalmia	-	-	-	Bil	Bil	Bil (L > R)	-	-	Bil
Coloboma	-	-	-	Bil	-	R	-	-	Bil
Cataract	-	-	-	-	-	R	-	-	-
Other	Lower lid cyst (R)	-	Ocular cyst (Bil)	RD and nodule near retina with small calcifications	-	-	-	-	Abnormal ASM
Craniofacial									
Nasal anomalies	-	-	-	-	-	-	EN	-	WNB, EN, SP
SPF	-	-	-	-	-	-	+	+	+
DCM	-	-	-	-	-	-	-	+	+
Other	-	-	-	-	-	-	-	-	Telecanthus, Micrognathia, EF, EP, EUL, HF, FC
Developmental									
ID	-	+	+	-	-	-	+	-	-
Motor Delay	-	+	+	-	-	-	+	-	-
Speech Delay	-	Nonverbal	+	-	-	-	-	-	-
Autism	-	+	Autistic features	-	-	-	-	-	-
MRI findings	Normal	NK	Normal	NK	Normal	NK	Rudimentary ON and chiasm	Hypoplastic ON, Bil DL	NK
Other findings	Fistula-in-ano	-	Seizures	-	Crowded teeth	-	-	-	-

Variables are reported according to GRCh37/hg19.

ASM anterior eye segment morphology, Bil bilateral, DCM downturned corners of mouth, DL dysplastic lens, EF epicanthic folds, EN enlarged nares, EP eyelid ptosis, EUL everted upper lip, F female, FC full cheeks, HC head circumference, HF high forehead, ID intellectual disability, L left, M male, m months, NK not known, ON optic nerves, R right, RD retinal detachment, SP short philtrum, SPF short palpebral fissures, WNB wide nasal bridge, y year.

^a(NM_000693.4).

The original article has been corrected.



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