

Additional File 2. Genetic status of ICF patients.

Patients (Gender)	Amino acid changes in DNMT3B	Amino acid changes in ZBTB24	Birth year	Follow-up Status ^(reference)
ICF1	NP_008823.1			
pG (M)	p. I41fsX42 / p.S780L		1985	Deceased age 16 ^[1]
Cor (F)	p. A603T / p.R807delinsSTP			NA ^[2]
pR (F)	p. D817G / p. D817G		1991	Necker Hospital (Paris, France) ^[3]
pW (F)	p.V613A / p.R826C		2004	NA ^[4]
pT (F)	p. D817G / p. D817G		2004	Deceased ^[5]
pI (M)	p. G663S / p.V726G		1972	Deceased ^[6]
pH (M)	p.V818M / p.V818M		1990	University Hospital Center (Nancy, France) ^[3]
P1*(F)	p. S655L / ?		2010	Necker Hospital (Paris, France)
pY*(M)	p.T775I / p.T775I		1996	Necker Hospital (Paris, France)
P2*(M)	p. R104X / p.I721T		2008	Necker Hospital (Paris, France)
P3^a*(F)	p. G583S / p. G583S		2009	Necker Hospital (Paris, France)
P4^a*(F)	p. G583S / p. G583S		2013	Necker Hospital (Paris, France)
P5 (M)	p. K770E / p. K770E		1991	(Lebanon) ^[7]
ICF2		NP_055612.2		
pP (F)		p.R457X / p.R457X	1981	NA ^[8]
P6^b (M)		p.H132Q fsX19 / p.H132Q fsX19	2003	Hotel-Dieu de France Hospital (Beirut, Lebanon) ^[9]
pD^b (M)		p.H132Q fsX19 / p.H132Q fsX19	1997	Hotel-Dieu de France Hospital (Beirut, Lebanon) ^[9]
pV^b (M)		p.H132Q fsX19 / p.H132Q fsX19	1998	Hotel-Dieu de France Hospital (Beirut, Lebanon) ^[9]
P7 (M)		p.R320X / p.R320X	1971	Deceased ^[10]
P8 (F)		p.K263X / p.C327W fsX54	1996	Necker Hospital (Paris, France) ^[10]
ICFX				
pC (F)				NA ^[3]
pS (F)			1978	Necker Hospital (Paris, France) ^[3]
pU (M)				NA
pN (F)				NA ^[3]

M = male ; F = female ; * and grey shade = newly described ICF1 patients in the current study ; ^{a, b} siblings; NA; not available

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