

# Mutations in *PRDM5* in Brittle Cornea Syndrome Identify a Pathway Regulating Extracellular Matrix Development and Maintenance

Emma M.M. Burkitt Wright, Helen L. Spencer, Sarah B. Daly, Forbes D.C. Manson, Leo A.H. Zeef, Jill Urquhart, Nicoletta Zoppi, Richard Bonshek, Ioannis Tosounidis, Meyammai Mohan, Colm Madden, Annabel Dodds, Kate E. Chandler, Siddharth Banka, Leon Au, Jill Clayton-Smith, Naz Khan, Leslie G. Biesecker, Meredith Wilson, Marianne Rohrbach, Marina Colombi, Cecilia Giunta, and Graeme C.M. Black\*

(American Journal of Human Genetics 88, 767–777; June 10, 2011).

In the original version of this paper, there was a misalignment of numbering of individuals of family BCS-001 in Table 1, affecting individuals IV:7 (unaffected), IV:8 (heterozygous), and IV:10 (homozygous). The corrected table appears below. The authors regret the error.

**Table 1. Phenotypic Characteristics of Individuals with *PRDM5* Mutations from Families BCS-001 and BCS-002**

	BCS-001					BCS-002									
	IV:4	IV:6	IV:9	IV:10	IV:8	V:2	V:3	V:5	V:6	IV:6	V:1	V:4	V:5	IV:3	
Homozygous/heterozygous	hom	hom	hom	hom	het	het	het	het	het	hom	hom	hom	hom	het	
Corneal rupture	+	+	+	+											
Myopia	+	+	+	+	+						+	+	+		
Blue sclera	+	+	+	+	+	+	+	+	+	+	+	+	+		
Keratoconus	+	+	+	+	+						+				
Keratoglobus	+	+	+	+											
Megalocornea															
Poor healing/abnormal scarring											+				
Soft skin/easy bruising	+	+	+	+											
Treatment for DDH	+										+	+			
Femoral epiphyseal changes		+	+	+											
Scoliosis	+														
Small joint hypermobility	+	+	+	+	+	+	+	+	+	+	+	+	+		
Fractures										+		+	+		
Myalgia	+	+								+					
Abnormal gait	+	+	+	+						+		+			
Deafness	+	+	+	+						+	+	+	+		
Hypercompliant TMs	+	+	+	+						+	+	+			
Other features	P									LD	H	LD	LD CLP PKU		
CCT less than 400 microns	+	+	+	+						+	+	+	+		
CCT 400 to 550 microns					+	+	+	+	+					+	

Affected, homozygous, individuals in each family are indicated. + indicates present; and empty box indicates not present. N/A indicates data not available. The following abbreviations are used: DDH, developmental dysplasia of the hip; TM, tympanic membrane; CCT, central corneal thickness. P, primiparous cervical incompetence; LD, learning disability; H, hernia (inguinal, umbilical or epigastric); CLP, cleft lip and palate; PKU, phenylketonuria.

\*Correspondence: [graeme.black@manchester.ac.uk](mailto:graeme.black@manchester.ac.uk) (G.C.M.B.)

DOI 10.1016/j.ajhg.2011.07.013. ©2011 by The American Society of Human Genetics. All rights reserved.