

Supplemental Information

A Recurrent *PDGFRB* Mutation

Causes Familial Infantile Myfibromatosis

Yee Him Cheung, Tenzin Gayden, Philippe M. Campeau, Charles A. LeDuc, Donna Russo, Van-Hung Nguyen, Jiancheng Guo, Ming Qi, Yanfang Guan, Steffen Albrecht, Brenda Moroz, Karen W. Eldin, James T. Lu, Jeremy Schwartzentruber, David Malkin, Albert M. Berghuis, Sherif Emil, Richard A. Gibbs, David L. Burk, Megan Vanstone, Brendan H. Lee, David Orchard, Kym M. Boycott, Wendy K. Chung, and Nada Jabado

Table S1. Clinical characteristics and *PDGFRB* mutations in four different families and five simplex cases with IM

SUBJECT	CLINICAL HISTORY	TISSUE	GERMLINE MUTATION c.1681C>T	SOMATIC MUTATION c.1998C>A
FAMILIAL CASES				
Family 1				
I-1	Solitary myofibroma at a young age that did not require further treatment.	Blood	Heterozygous	Wild type
I-2	Unaffected	Blood	Wild type	Wild type
II-1	Diagnosed at the age of 4 years with multiple fibromas of the skin, the majority of which spontaneously regressed.	Blood	Heterozygous	Wild type
II-2	Referred at 6 month of age with multiple myofibromas of the skin and visceral myofibroma of the orbit and supra-nasal region.	Blood/Tumor	Heterozygous	Wild type
Family 2				
I-1	Unaffected	Colon tissue	Wild type	Wild type
I-2	Unaffected	Blood	Wild type	Wild type
II-2	Multiple subcutaneous on the cheeks, shoulder, back, abdominal wall	Blood	Heterozygous	Wild type
III-1	Mutiple myofibromas in first year of life	Tumor – abdominal wall	Heterozygous	Heterozygous
		Tumor – left upper gingival border	Heterozygous	Heterozygous

		Tumor – left neck mass	Heterozygous	Wild type
		Blood	Heterozygous	Wild type
Family 3				
I-1	Unaffected	Blood	Wild type	Wild type
I-2	Unaffected	Blood	Wild type	Wild type
II-1	Multiple myofibromas that spontaneously resolved	Blood	Heterozygous	Wild type
II-2	Unaffected	Blood	Wild type	Wild type
III-1	Unaffected	Blood	Wild type	Wild type
III-2	Multiple myofibromas of the skin of the face and upper arms that spontaneously resolved at the age of 4 years	Blood	Heterozygous	Wild type
III-3	Multiple myofibromas of the skin of the face and upper arms that spontaneously resolved at the age of 4 years	Blood	Heterozygous	Wild type
Family 4				
I-1	Unaffected	Blood	Wild type	Wild type
I-2	Left neck swelling at 7 months of age, which was subsequently excised and histopathology revealed fibromatosis	Blood	Heterozygous	Wild type
II-1	Visceral myofibroma of the abdominal wall and an ulcerated lesion over the left fifth metacarpophalangeal joint at 3 weeks of age	Blood	Heterozygous	Wild type
II-2	Unaffected	Blood	Wild type	Wild type
II-3	Referred at 11 weeks of age with three tumors involving the scalp and trunk that subsequently regressed	Blood	Heterozygous	Wild type
SIMPLEX CASES				
Simplex 1	Myofibroma of the cheek at birth	Tumor	Wild type	Wild type
Simplex 2	Pelvic myofibroma resected at 34 days of age	Tumor	Wild type	Wild type
Simplex 3	Diagnosed at 13 days of age with large lesion originating from the right hemi-diaphragm	Blood/Tumor	Wild type	Wild type
Simplex 4	Visceral myofibroma at 5 days of age	Tumor	Wild type	Wild type
Simplex 5	Myofibromas of femur and lung at 1 month age	Blood/Tumor	Wild type	Wild type

Table S2. PCR primer sequences used to validate c.1681C>T and c.1998C>A mutations in *PDGFRB*.

Mutation	Primer sequence		Amplicon length
c.1681C>T (p.Arg561Cys)	Forward	5'-TGTCCTAGACGGACGAACCT-3'	274 bp
	Reverse	5'-CAGAGAGTCTTCCCACCCAA-3'	
c.1998C>A (p.Asn666Lys)	Forward	5'-AGAATAGGCTCCTGTGGTGG-3'	289 bp
	Reverse	5'-AGCAGGAGTGTGCTGTTGTG-3'	