Supplementary Methods

LightScanner Melt Curve Analysis

PCRs for LightScanner melt curve analysis consisted of 10ng of template DNA, and final concentrations of 0.25µM of each primer, 1x HotShot Diamond Master Mix (ClentLife) and 1x LCGreen Plus Dye (BioFire Diagnostics Inc) in a total volume of 10µl. PCRs were typically one cycle of 5 minutes 95^{o}C, 45 cycles of 95^{o}C 30s, annealing temperature 30s and 72^{o}C 45s, followed by a final denaturation step of 95^{o}C for 30s. PCRs were performed in Framestar® 96 (4titude) 96-well plates with a 20µl mineral oil overlay. Melt curves were generated on a LightScanner® (Idaho Technology Inc) using autoexposure, a starting temperature of 75^{o}C and a stop temperature of 98^{o}C. Data was analysed using the LightScanner® Instrument & Analysis Software (Idaho Technology Inc). Melt curves were normalised prior and post the major melt transition and aberrant curves detected using the autogroup function and manual inspection. Samples with an initial fluorescence of less than 600 were excluded from analysis, while those with a starting fluorescence less than 800 were not included during normalisation.

Supplementary Table 1: Primers used for Screening *HMX1*

Exon	Primer	Sequence	PCR	Condition
1	OLFM2_ Ex1F	CAAGCCAGAGAGTGCACGTC	419	60°C, 10% DMSO, 45x
	OLFM2_ Ex1R	gcaacaaagactcggagcga		
1st alternative exon 1	OLFM2_alt1Ex1F	GAAGCACAGGGGTAGAGGG	461	TD, 10% DMSO, 45x
	OLFM2_ alt1Ex1R	TTATAAGAGGAGCCCGCCAG		
2	OLFM2_ Ex2F	CTGGAGAGGAGCTGGATTATCA	388	55°C, 45x
	OLFM2_ Ex2R	CATCTGTGGTTTCCTTGGGC		
3	OLFM2_ Ex3F	TGTGGCTCATATTGGACCCT	336	50°C, 45x
	OLFM2_ Ex3R	AGCTCTTGTCTGTGGCATCT		
4	OLFM2_ Ex4F	GCTTCTAGGCACAAACAGGT	377	60°C, 45x
	OLFM2_ Ex4R	TGAGTCAGAGGTTGGAGTCA		
5	OLFM2_ Ex5F	TCCAGGACACTTTGGGCTAC	295	55°C, 35x
	OLFM2_ Ex5R	CTTCATCTTTGCCTGGCCTC		
6	OLFM2_ Ex6F1	ACAGGCAGAATGAAAAGGGC	300	62°C, 35x
	OLFM2_ Ex6R1	TGCTCTACGTGACCAACTCC		
	OLFM2_ Ex6F2	ACGTCCGTGTACTCGTAACT	372	62°C, 45x
	OLFM2_ Ex6R2	GAGCAACGTGGTGGTCAAAT		
	OLFM2_ Ex6F3	GGAGTAGGGGAAGGTGTTGT	386	50°C, 45x
	OLFM2_ Ex6R3	GTCAACAGAGTTCCCATGACT		

Note: we were unable to amplify the second alternative exon 1

TD = Touchdown PCR from 70°C to 55°C, decreasing by 1°C per cycle

Supplementary Table 2: Primers used to confirm *OLFM2* expression in cDNA

Primer	Sequence	PCR	Condition
Forward	CACATGACGCGCCCCTAG	190	55°C, 10% DMSO, 35x
Reverse	gcaacaaagactcggagcga		

Note: The Ex6F2/R2 primer pair (Supplementary Table 1) were also used to confirm expression.