

**Supplemental Table S1. Characteristics of children with IBMFS and low bone mineral density (BMD HAZ  $\leq$ -2).**

UPN	Diagnosis	Age, yrs	Sex	Height Z-score	BMD HAZ	Treatment	Transfusion History	HSCT	Fracture History	Endocrine; Hypogonadism	Bisphosphonate
NCI 2-1	DBA	14	M	-3.24	FN: -2.39 LS: 0.55	Prednisone x 9 yrs	PRBC x 12 years; iron overload	No	No	not tested	Alendronate x 2 yrs
NCI 204-1	DC	12	M	-1.15	FN: -4.27 LS: -0.65	Prednisone high-dose x 1 yr for pulmonary fibrosis	No	Age 4 yrs; HLA-ID sibling	No	No	No
NCI 154-1	DC	15	M	-0.71	FN: -2.01 LS: -1.05	Oxymetholone; Androderm x 1 yr	PRBC x 2 years; Iron overload	No	No	No	No
NCI 220-1	DC	15	M	0.82	FN: -2.62 LS: -0.31	None	None	No	No	No	No
NCI 194-1	DC-like*	17	F	0.51	FN: -2.55 LS: -2.71	Oxymetholone x 1 yr; glucocorticoid premedication x 3 yrs	PRBC and platelets x 3 yrs; iron overload	No		No	No
NCI 291-1	DC	19	M	-1.6	FN: -2.17 LS: -1.37	None	None	No	No	No	
NCI 285-2	DC-like*	19	M	-0.38	FN: -2.52 LS: -0.22	Prednisolone and oxymetholone x 10 yrs	No	No	No	Low testosterone	No
NCI 243-1	FA	13	M	-0.21	FN: -2.00 LS: 0.32	Levothyroxine; growth hormone therapy	No	Age 9 yrs; MUD	No	Hypothyroid; Growth hormone deficiency	No
NCI 241-1	FA	16	M	-3.2	FN: -2.04	Levothyroxine	No	Age 6 yrs; HLA-ID	No	Hypogonadism and hypothyroid	No

					LS: -0.72			sibling			
NCI 75-1	FA	18	M	-0.89	FN: -0.76 LS: 1.51	Oxymetholone x 4 yrs; prednisone x 5 yrs	No	No	Vertebral compression fractures age 14	DXA age 14: Osteoporosis	Alendronate x 2 yrs
NCI 173-1	SDS	12	M	-6.63	FN: -2.14 LS: -0.02	None	No	No	No	No	No

\*The diagnosis was subsequently changed to DC-like in these 2 patients with very short telomeres because they did not meet the clinical criteria for the diagnosis of DC and no known mutated gene was identified.

FN, femoral neck. LS Lumbar spine. HAZ, height-adjusted Z-score. PRBC, packed red blood cells. HLA-ID, HLA identical HSCT, hematopoietic stem cell transplantation. MUD, matched unrelated donor. UPN, unique patient identifier. NCI, National Cancer Institute

**Supplemental Table S2. Characteristics of adults with IBMFS and low bone mineral density (BMD Z-score  $\leq$ -2).**

UPN	Diagnosis	Age	Sex	Height Z-score	BMD Z-score	Treatment	Transfusions	HSCT	Fractures	Hypogonadism	Bisphosphonate
NCI 193-5	DBA	26	M	-1.37	FN: -2.0 LS: -1.4	Prednisone X 10+ yrs	PRBC x 26 yrs; iron overload	No	AVN hip	Yes	
NCI 57-1	DBA	30	F	-0.52	FN: -1.4 LS: -0.8	estrogen/progesterone; insulin	PRBC x 18+ yrs; iron overload	No	No	POI	Alendronate x 7 yrs
NCI 167-1	DC	29	M	-0.82	FN: -2.20 LS: -3.90	G-CSF x 2 yrs; AML Rx; insulin for type 1 diabetes 27 yrs	PRBC x 3 yrs; iron overload	No	No	No	
NCI 26-1	DC	29	M	-1.93	FN: NA LS: -2.20	Warfarin for hyperthrombotic state	No	No	AVN hip	No	No
NCI 9-1	FA	42	F	-1.21	FN: -1.9 LS: -4.0	Prednisone X 10+ years	No	No	No	POI with premature menopause age 34	No
NCI 144-1	FA	37	F	-3.12	FN: -1.9 LS: -2.4	Nandronolone	No	No	No	POI with premature menopause age 34	Alendronate x 2 yrs
NCI 73-2	FA	27	F	-2.45	FN: -1.9 LS: -2.5	None	No	No	No	POI	No
NCI 169-1	FA	23	F	-2.5	LS: -4.7: FN: -4.5	None	No	Age 7; HLA-ID sibling	vertebral compression fractures	POI	Alendronate x 2 yrs
NCI 7-1	FA	52	F	-1.4	FN: -0.7	Estrogen x 15 yrs	No	No	No	Premature menopause age 35	Alendronate x 10 yrs

					LS: -0.2;						
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FN, femoral neck. LS Lumbar spine. HAZ, height-adjusted Z-score. PRBC, packed red blood cells. HLA-ID, HLA identical. HSCT, hematopoietic stem cell transplantation. MUD, matched unrelated donor. UPN, unique patient identifier. NCI, National Cancer Institute

Supplemental Figure S1: Correlation of spine and femoral neck BMD Z-score and BMaD with height in adults with IBMFS

