

Telomere length in inherited bone marrow failure syndromes

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ALTER ET AL: SUPPLEMENTARY APPENDIX

Supplementary Table S1: Telomere Lengths below First Percentile in non-DC Patients compared with DC Patients

		DC patients vs non-DC patients						
	Cell type	N abnl/total	OR	95% CI	Sens (%)	Spec (%)	PPV (%)	NPV (%)
DC	Granulocytes	89/95						
	Lymphocytes	89/100						
	CD45+ naïve T cells	85/99						
	CD45- memory T cells	85/99						
	CD20+ B cells	79/93						
	CD57+ NK/NKT cells	73/93						
	≥4 cell types	86/100						
	>3 cell types*	85/99						
DBA	Granulocytes	7/33	0.02	0.005-0.07	21	6	7	19
	Lymphocytes	1/34	0.004	0.0001-0.02	3	11	1	25
	CD45+ naïve T cells	1/34	0.005	0.001-0.04	3	14	1	30
	CD45- memory T cells	2/34	0.01	0.001-0.05	6	14	2	30
	CD20+ B cells	1/34	0.005	0.001-0.04	3	15	1	30
	CD57+ NK/NKT cells	0/31	0	0-0.04	0	22	0	39
	≥4 cell types	1/34	0.005	0.001-0.04	3	14	1	30
	≥3 cell types*	1/34	0.005	0.001-0.04	3	14	1	30
FA	Granulocytes	14/28	0.07	0.02-0.2	50	6	14	30
	Lymphocytes	5/30	0.02	0.006-0.09	17	11	5	31
	CD45+ naïve T cells	5/30	0.03	0.009-0.1	17	14	6	36
	CD45- memory T cells	3/30	0.007	0.003-0.07	10	14	3	34
	CD20+ B cells	6/24	0.04	0.02-0.2	25	15	7	44
	CD57+ NK/NKT cells	2/29	0.04	0.002-0.1	7	22	3	43
	≥4 cell types	3/30	0.01	0.003-0.07	10	14	3	34
	≥3 cell types*	2/30	0.01	0.001-0.06	7	14	2	33
SDS	Granulocytes	3/14	0.02	0.003-0.1	7	21	6	3
	Lymphocytes	1/14	0.01	0.0002-0.08	7	11	1	46

	CD45+ naïve T cells	1/14	0.01	0.0003-0.1	7	14	1	52
	CD45- memory T cells	2/14	0.03	0.003-0.1	14	14	2	54
	CD20+ B cells	2/14	0.03	0.003-0.2	14	15	3	54
	CD57+ NK/NKT cells	1/14	0.02	0.0005-0.2	7	22	1	61
	≥4 cell types	1/14	0.01	0.0003-0.1	7	14	1	52
	≥3 cell types*	1/14	0.01	0.0003-0.1	7	14	1	52

Comparison of numbers with TL abnormal (below first percentile) among DC and non-DC patients. OR, odds ratio for having DC. CI, 95% confidence interval; sens, sensitivity; spec, specificity; PPV, positive predictive value; and NPN, negative predictive value. Denominators varied because some cell types did not have sufficient cells. *Excluding granulocytes and NK cells.

Supplementary Table S2: Telomere Length Z-scores below -2.326 in non-DC Patients compared with DC Patients

Non-DC	Cell type	N abnl/total	DC patients vs non-DC patients					
			OR	95% CI	Sens (%)	Spec (%)	PPV (%)	NPV (%)
	Granulocytes	88/95						
	Lymphocytes	90/100						
	CD45+ naïve T cells	83/99						
	CD45- memory T cells	84/99						
	CD20+ B cells	78/93						
	CD57+ NK/NKT cells	70/93						
	≥4 cell types	86/100						
	>3 cell types*	85/99						
DBA	Granulocytes	7/33	0.03	0.008-0.09	21	10	8	26
	Lymphocytes	2/34	0.01	0.001-0.05	6	14	2	30
	CD45+ naïve T cells	2/34	0.01	0.001-0.06	6	17	2	35
	CD45- memory T cells	2/34	0.01	0.001-0.06	6	16	2	33
	CD20+ B cells	1/34	0.007	0.0002-0.05	3	18	1	34
	CD57+ NK/NKT cells	1/31	0.02	0.0004-0.1	3	32	2	50
	≥4 cell types	1/34	0.006	0.0001-0.04	3	16	1	33
	≥3 cell types*	1/34	0.005	0.0001-0.04	3	14	1	30
FA	Granulocytes	13/28	0.09	0.03-0.3	46	10	13	38
	Lymphocytes	4/30	0.03	0.005-0.09	13	14	4	35
	CD45+ naïve T cells	6/30	0.05	0.02-0.2	20	17	7	42
	CD45- memory T cells	3/30	0.02	0.004-0.08	10	16	4	37
	CD20+ B cells	6/24	0.07	0.02-0.2	25	18	7	49
	CD57+ NK/NKT cells	2/29	0.04	0.004-0.2	7	32	3	53
	≥4 cell types	3/30	0.02	0.004-0.08	10	16	3	37
	≥3 cell types*	2/30	0.01	0.001-0.06	7	14	2	33
SDS	Granulocytes	3/14	0.03	0.005-0.1	21	10	3	45
	Lymphocytes	2/14	0.03	0.003-0.1	14	14	2	54

	CD45+ naïve T cells	1/14	0.02	0.0004-0.1	7	17	1	57
	CD45- memory T cells	1/14	0.02	0.0003-0.1	7	16	1	55
	CD20+ B cells	2/14	0.04	0.004-0.2	14	18	3	59
	CD57+ NK/NKT cells	1/14	0.04	0.0008-0.3	7	32	2	70
	≥4 cell types	2/14	0.03	0.003-0.2	14	16	2	57
	≥3 cell types*	2/14	0.03	0.003-0.2	14	14	2	54

Abbreviations defined in Table 3

Supplementary Figure S1: Telomere length Z-score boxplots in patients with DC, FA, DBA, and SDS. Dashed lines represent the mean and +2.326 or -2.326 standard deviations (SD) from the mean (equivalent to the first percentile) for normal individuals. Top panels: Granulocytes, lymphocytes, and CD45RA-positive/CD20-negative naïve T-cells. Bottom panels: CD45RA-negative memory T-cells, CD20-positive B-cells, and total CD57-positive NK cells. The + sign indicates outliers.

Supplementary Figure S2: Telomere length Z-score boxplots in lymphocytes from patients with DC, FA, DBA, and SDS according to severity of bone marrow failure. Dashed lines represent the mean and +2.326 or -2.326 standard deviations (SD) from the mean (equivalent to the first percentile) for normal individuals. X-axes: None (no aplastic anemia), MAA (moderate aplastic anemia) and SAA (severe aplastic anemia).



