Expanding the clinical phenotype of autosomal dominant dyskeratosis congenita caused by *TERT* mutations

Lina Basel-Vanagaite,[1,2] Inderjeet Dokal,[3] Hannah Tamary,[2,4] Abraham Avigdor,[5] Ben Zion Garty,[3,6] Alexander Volkov,[7] and Tom Vulliamy[3]

[1]Schneider Children's Medical Center of Israel and Raphael Recanati Genetics Institute, Rabin Medical Center, Beilinson Campus, Petah Tikva, Israel; [2]Sackler Faculty of Medicine, Tel Aviv University, Tel Aviv, Israel; [3]Academic Unit of Paediatrics, Institute of Cell and Molecular Science, Barts and the London School of Medicine and Dentistry, UK; [4]Pediatric Hematology-Oncology, Schneider Children's Medical Center of Israel, Petah Tikva, Israel; [5]Division of Hematology and Bone Marrow Transplantation, Chaim Sheba Medical Center, Tel-Hashomer, Israel; [6]Department of Pediatrics B, Schneider Children's Medical Center of Israel, Petah Tikva, Israel; [7]The Department of Pathology, Chaim Sheba Medical Center, Tel-Hashomer, Israel

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Supplementary Table 1. Clinical features of the known or putative mutation carriers.

Individual	*Current age/age at onset of the disease (yr)*	*CBC (RBC×10¹²/L; WBC Plt/10⁹/L, Hb-g%, MCV-μm³)*	*Bone marrow examination*	*Liver and spleen abnormalities (GOT-IU/L, GPT-IU/L, bilirubin-mg/dL)*	*Pulmonary symptoms*	*Premature grey hair*	*Other*	*Cause of death*
I-1	deceased/57	WBC 4.9 Hb 11 Plt 26	Hypocellularity, dyserythropoiesis	Liver enzymes NA, bilirubin 2.5	None	NA	–	Myocardial infarction
II-1	deceased/35	NA, abnormal	NA	NA	Yes	Yes	–	Respiratory failure
II-4	deceased/24	NA, abnormal	NA	Liver disease GOT-19 Bilirubin - 1.3 Splenomegaly	Yes	Yes	Osteoporosis, L1 vertebral fracture	Hepatic failure
II-7*	52/healthy	WBC 5.32 RBC 4.86 Hb 13.9 MCV 84.5 Plt 191	NP	GOT-16 GPT-14 Bilirubin-0.8	None	Yes	–	–
III-2*	30/ healthy	WBC 4.35 RBC 3.88 Hb 11.6 MCV 91 Plt 169	NP	GOT-19 GPT-17 Bilirubin 0.6	None	Yes	–	–
III-4	deceased/7	WBC 1.87 RBC 3.35 Hb 10.7 MCV 114 Plt 43	BM cellularity 25-30%; BM colony assay: markedly reduced BFU and CFU number and mega colonies	Liver disease with varicosis of portal veins Splenomegaly	Restrictive interstitial pulmonary disease	Yes	Retinal detachment at birth; Strabismus Cardiac fibrosis	Respiratory failure
III-6*	33/18	WBC 1.69 RBC 3.91 Hb 13.3 MCV 92 Plt 42	NA	GOT 46 GPT 163 CT–irregular liver texture Splenomegaly	CT lungs fibrotic changes in both apical regions	Yes	–	
III-7*	30/16	WBC 2.67 RBC 3.7 Hb 12.2 MCV 95 Plt 41	NA	GOT 31 GPT 27 Bilirubin 1.7 Splenomegaly	None	Yes	Dilated cardiomyopathy	

**: those shown to be heterozygous for TERT c.1892G>A. Samples from the other affected individuals were not available for genotyping. NA : not available; NP; not performed; PM: post mortem examination; GOT: glutamic oxaloacetate transaminase; GPT : glutamic pyruvate transaminase; BM: bone marrow; BFU: blood forming units; CFU: colony-forming units.*