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| **Name** | Sharon A. Savage, MD |
| **Current Position & Affiliation** | Clinical Director, Division of Cancer Epidemiology and Genetics, National Cancer Institute |
| **Country** | USA |
| **Major Field** | Inherited bone marrow failure syndromes |

## Educational Background

1991 - Bachelor of Science (BS) in Biochemistry: Worcester Polytechnic Institute, Worcester, MA, USA
1997 - Medical Doctor (MD) from the University of Vermont College of Medicine, Burlington VT, USA, in 1997
1997-2000 - Pediatrics Residency, Children's National Medical Center, Washington, DC,
2000-2003 - Pediatric Hematology-Oncology Fellowship, National Cancer Institute and Johns Hopkins University
2003-2006 - Clinical Research Fellow, National Cancer Institute

## Professional Experience

2019 – present Clinical Director: DCEG, NCI, NIH, Rockville, MD
2013 – present Branch Chief: Clinical Genetics Branch, DCEG, NCI, NIH, Rockville, MD
2012 – present Senior Investigator (tenured): Clinical Genetics Branch, DCEG, NCI, NIH
2006 – 2012 Investigator, Tenure-Track: Clinical Genetics Branch, Division of Cancer Epidemiology and Genetics (DCEG), NCI, NIH, Rockville, MD
2005 – 2011 Attending Physician: Pediatric Oncology Branch NCI, NIH, Bethesda, MD

## Other Experience and Professional Memberships

*Selected:*
Member (elected 2013), American Society for Clinical Investigation
Member, American Society of Hematology (ASH)
ASH Scientific Committee on Bone Marrow Failure: Member (2021-2025), Vice Chair (2024), Chair (2025)
Associate Editor for Molecular Biology, British Journal of Haematology, 2023 to present

## Main Scientific Publications

**Major scientific publications since 2020**. Complete List in MyBibliography: https://www.ncbi.nlm.nih.gov/myncbi/sharon.savage.1/bibliography/public/

**Telomere Biology:**


- Raj HA, Lai T-P, Niewisch MR, Giri N, Wang Y, Spellman SR, Aviv A, Gadalla SM, Savage SA: The distribution and accumulation of the shortest telomeres in telomere biology disorders. British J Haematol 2023;203(5):820-828. PMID: 37354000 PMC10748793
- Niewisch MR, Giri N, McReynolds LJ, Alsaggaf R, Bhala S, Alter BP, Savage SA: Disease progression and clinical outcomes in telomere biology disorders. Blood 2022;139(12):1807-1819 PMID: 34852175 PMC8952184

- Niewisch MR, Kim J, Giri N, Lunger JC, McReynolds LJ, Savage SA: Genotype and associated cancer risk in patients with telomere biology disorders. JAMA Network Open 2024;7(12):e2450111 PMID: 39661387 PMC11635530
- *Gutierrez-Rodrigues F, *Groarke EM, *Thongon N, Rodriguez-Sevilla JJ, Catto LFB, Niewisch MR, Shalhoub R, McReynolds LJ, Cle DV, Patel BA, Ma X, Hironaka D, Donaires FS, Spitofsky N, Santana BA, Lai T, Alemu L, Kajigaya S, Darden I, Zhou W, Browne PV, Paul S, Lack J, Young DJ, DiNardo CD, Aviv A, Ma F, de Oliveria MM, de Azambuja AP, Dunbar CE, Giri N, Alter BP, Bomfim C, Wu CO, Garcia-Manero G, Savage SA, **Young NS, **Calado RT, **Colla S: Clonal landscape and clinical outcomes of telomere biology disorders: somatic rescuing and cancer mutations. Blood 2024;144(23):2402-2416. PMID: 39316766 PMC11862815
- de Andrade KC, Pinto EM, Zhao T, Zeigler LP, Kim J, Giri N, Haley JS, McReynolds LJ, Florez-Vargas O, Phillips AH, Kriwacki RW, Akinniyi SA, Cohen SB, Emerson MR, Smelser DT, Urban GM, Fridman C, Zambetti GP, Bryan TM, Carey DJ, Garcia CK, Stewart DR, Savage SA: *TERT* c.3150G>C (p.K1050N): a founder Ashkenazi Jewish variant associated with telomere biology disorders. npj Genomic Med 2025;10:46 PMID: 40456748 PMC12130525

**Inherited Bone Marrow Failure Syndromes (IBMFS)**

- McReynolds LJ, Rafati M, Wang Y, Ballew BJ, Kim J, Williams VV, Zhou W, Hendricks RM, Dagnall C, Freedman ND, Carter B, Strollo S, Hicks B, Zhu B, Jones K, Paczesny S, Marsh SGE, Spellman SR, He Meilun, Wang T, Lee SJ, Savage SA*, Gadalla SM*: Genetic testing in severe aplastic anemia is required for optimal hematopoietic cell transplant outcomes. Blood 2022;140(8):909-921 *equal contributions. PMID: 35776903; PMCID: PMC9412004
- Gianferante DM, Mendez KJW, Cole S, Gadalla SM, Alter BP, Savage SA, Giri N: Genotype-phenotype associations in individuals with Diamond Blackfan anemia. EJHaem 2024:5(6):1117-1124 PMID: 39691264 PMC11647742

**Li-Fraumeni Syndrome (LFS)**:

- Fortuno C, Lee K, Olivier M, Pesaran T, Mai PL, de Andrade KC, Attardi LD, Crowley S, Evans DG, Feng B, Major Foreman AK, Frone MN, Huether R, James PA, McGoldrick K, Mester J, Seifert BA, Slavin TP, Witkowski L, Zhang L, Plon SE, Spurdle AB, Savage SA, on behalf of the ClinGen TP53 Variant Curation Expert Panel: Specifications of the ACMG/AMP variant interpretation guidelines for germline TP53 variants. Human Mutat 2020;42(3):223-236 PMID:33300245
- de Andrade KC*, Khincha PP*, Hatton JN, Frone MN, Wegman-Ostrosky T, Mai PL, Best, AF, Savage SA: Cancer incidence, patterns, and genotype-phenotype associations in an observational study of individuals with pathogenic or likely pathogenic germline TP53 variants. Lancet Oncology 2021;22(12):1787-1798 PMID: 34780712
- de Andrade KC, Strande NT, Kim J, Haley JS, Hatton JN, Frone MN, Khincha PP, Thone GM, Mirshahi UL, Schneider C, Desai H, Dove JT, Smelser DT, Penn Medicine BioBank, Regeneron Genetics Center, Levine AJ, Maxwell KN, Stewart DR, Carey DJ, Savage SA: Genome-first approach to characterize the prevalence and cancer phenotypes of pathogenic or likely pathogenic germline TP53 variants. HGG Adv 2023;5(1):100242 PMID: 37777824 PMC10589747