BMC Genomics

CORRECTION

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Correction: Candidate variants in DNA replication and repair genes in early-onset renal cell carcinoma patients referred for germline testing

Elena V. Demidova[1,2], Ilya G. Serebriiskii[2,3], Ramilia Vlasenkova[2,3], Simon Kelow[4], Mark D. Andrake[3], Tiffiney R. Hartman[1,5], Tatiana Kent[6], James Virtucio[7], Gail L. Rosen[7], Richard T. Pomerantz[6], Roland L. Dunbrack Jr.[3], Erica A. Golemis[3,8], Michael J. Hall[1,9], David Y. T. Chen[10], Mary B. Daly[1,9*] and Sanjeevani Arora[1,11*]

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Following publication of the original article [1], it was reported that supplementary tables 1, 2 and 8 were missing from the published article. Additionally, the incorrect versions of Figs. 1 and 4 were published. The updated figures and supplementary files are included in this Correction and the original article has been updated.

Supplementary Information

The online version contains supplementary material available at https://doi.org/10.1186/s12864-023-09486-z.

Additional file 1: Supplementary table 1. List of candidate genes for WES analysis. **Supplementary table 2.** Annotation of candidate variants identified in the 22 eoRCC patients.

Additional file 2: Supplementary Table 8.

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Reference

1. Demidova EV, Serebriiskii IG, Vlasenkova R, et al. Candidate variants in DNA replication and repair genes in early-onset renal cell carcinoma patients referred for germline testing. BMC Genomics. 2023;24:212. https://doi.org/10.1186/s12864-023-09310-8.

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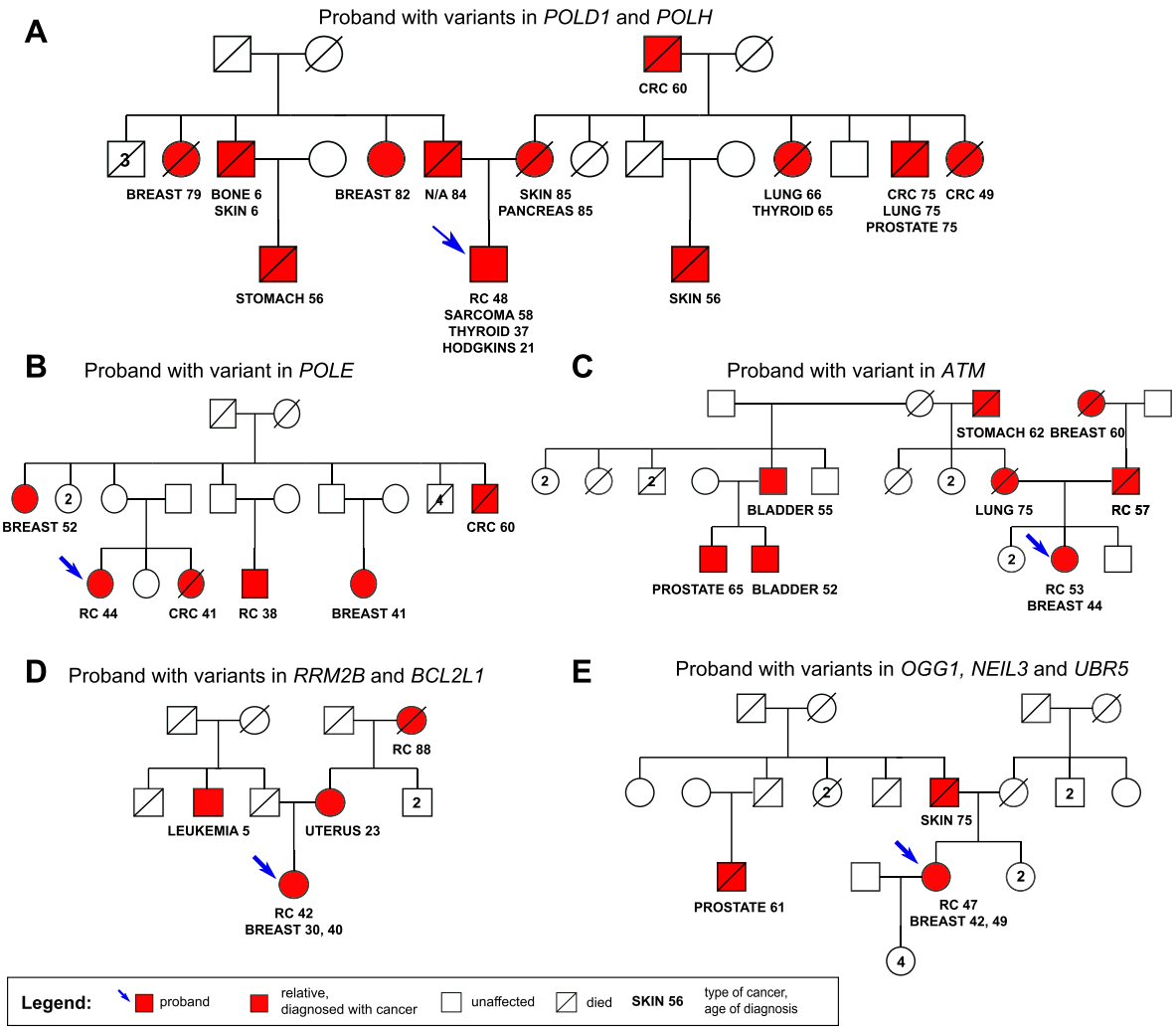
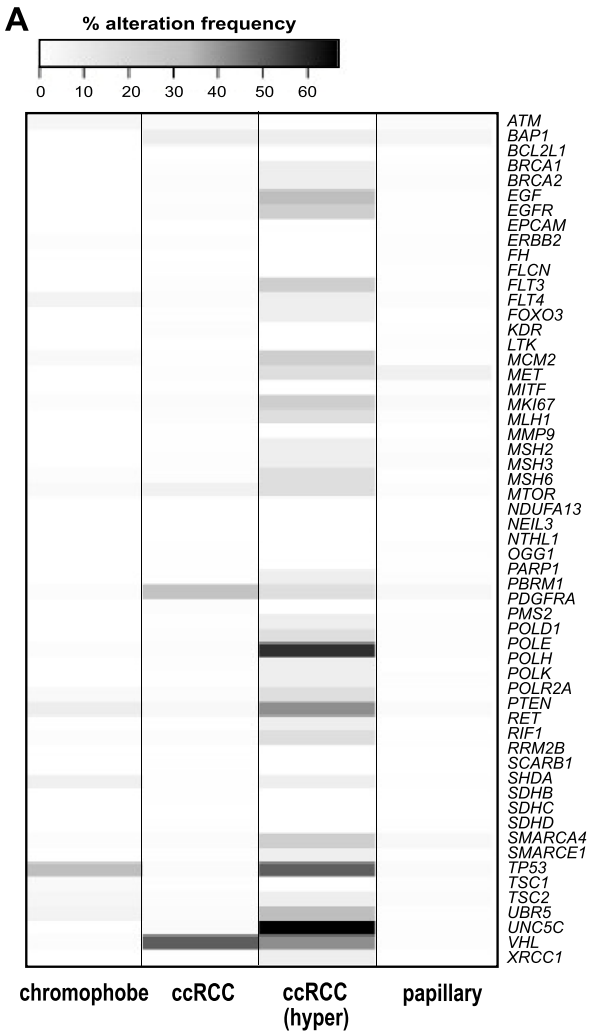
*Correspondence:
Mary B. Daly
Mary.Daly@fccc.edu
Sanjeevani Arora
Sanjeevani.Arora@fccc.edu
[1] Cancer Prevention and Control Program, Fox Chase Cancer Center, 333 Cottman Avenue, Philadelphia, PA 19111, USA
[2] Kazan Federal University, Kazan 420008, Russia
[3] Program in Cancer Signaling and Microenvironment, Fox Chase Cancer Center, Philadelphia, PA 19111, USA
[4] Department of Biochemistry and Molecular Biophysics, University of Pennsylvania, Philadelphia, PA 19104, USA
[5] Arcadia University, Glenside, PA, USA
[6] Department of Biochemistry & Molecular Biology, Sidney Kimmel Cancer Center, Thomas Jeferson University, Philadelphia, PA 19107, USA
[7] Ecological and Evolutionary Signal-Processing and Informatics Laboratory, Department of Electrical and Computer Engineering, College of Engineering, Drexel University, Philadelphia, PA 19104, USA
[8] Department of Cancer and Cellular Biology, Lewis Katz School of Medicine, Philadelphia, PA 19140, USA
[9] Department of Clinical Genetics, Fox Chase Cancer Center, 333 Cottman Avenue, Philadelphia, PA 19111, USA
[10] Department of Surgical Oncology, Fox Chase Cancer Center, Philadelphia, PA 19111, USA
[11] Department of Radiation Oncology, Fox Chase Cancer Center, Philadelphia, PA 19111, USA



F

POLD1	2	DNA polymerases
POLE	1	
POLH	1	
POLK	1	
ATM	4	Genes defective in diseases associated with sensitivity to DNA damaging agents
BLM	2	
RECQL4	6	
WRN	3	
RRM2B	1	Modulation of nucleotide pools
MPG	1	Base excision repair (BER)
MUTYH	1	
NEIL3	2	
NTHL1	1	
OGG1	3	
XRCC1	1	Other BER and strand break joining factors
PAPR1	1	PARP enzymes that bind to DNA
MLH1	1	Mismatch excision repair (MMR)
MLH3	1	
MSH2	1	
MSH3	3	
PMS1	1	
DCLRE1C	2	Non-homologous end-joining (NHEJ)
APTX	1	Editing and processing nucleases
HLTF	1	Ubiquitination and modification
ERCC2	1	Nucleotide excision repair (NER)
ERCC3	2	
ERCC4	2	
XPA	1	
XPC	1	
ERCC6	2	
BRCA1	2	Homologous recombination (HR)
GEN1	1	
MUS81	1	
RAD50	2	
RAD51D	1	
RAD54L	1	
BRCA2	4	Fanconi anemia (FA)
FANCA	2	
FANCB	1	
FANCD2	4	
FANCI	1	
FANCM	1	
PALB2	1	
DCLRE1A	1	Other identified genes with known or suspected DNA repair function
RECQL5	5	
ATR	2	Other conserved DNA damage response genes
RIF1	2	

Fig. 1 Select pedigrees from the eoRCC patient cohort and enrichment of predicted pathogenic variants in DNA repair genes in the cohort. **A**-**E**. Pedigrees of eoRCC patients with variants in: **A**—POLD1 and POLH; **B**—POLE; **C**—ATM; **D**—RRM2B and BCL2L1; **E**—OGG1, NEIL3 and UBR5. **F**. Summary of variants in genes and pathways, identifed in the cohort. In color—number of variants identifed for each gene. For detailed information, see Supplementary tables 1 and 2



B

Patient #	Tumor variants	TMB, mut/MB	MSS
P1	Pold1 V759I, Polh G209V	12.85	+
P2	Pole W1624X	14.44	+

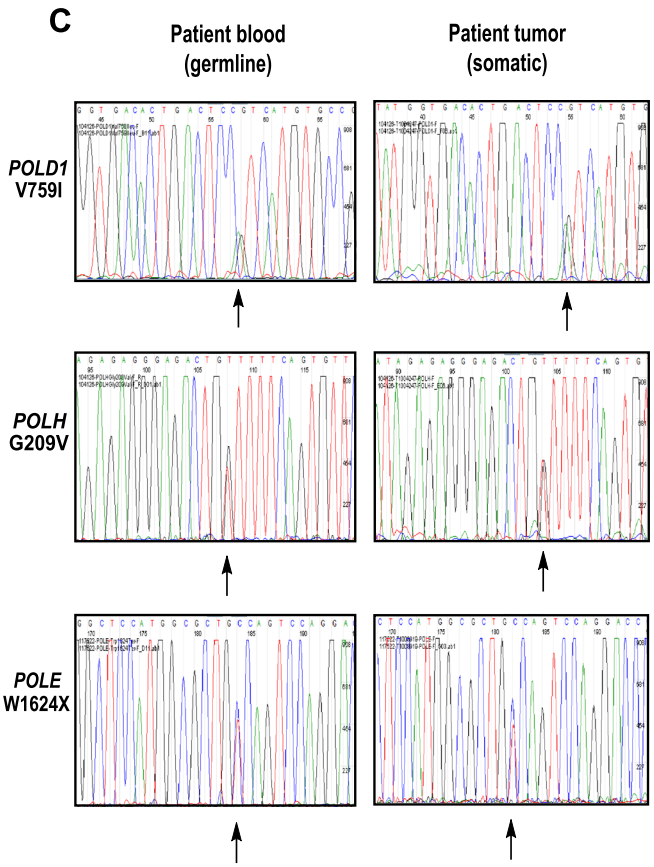


Fig. 4 Renal tumors carrying polymerase variants showed high TMB, MSS, and no LOH. **A**. Percent alteration frequency in 897 tumors from TCGA in diferent histological types of RCC: chromophobe ($n=66$), ccRCC—clear cell renal cell carcinoma ($n=538$), ccRCC (hyper)—hypermutated samples ($n=12$), papillary ($n=293$). **B**. TMB and MSS data are presented for Pt #1 (POLD1 V759I, POLH G209V) and Pt #2 (POLE W1624X). **C**. Tumor and normal Sanger sequencing for variants in Pt #1 (POLD1 V759I, POLH G209V) and Pt #2 (POLE W1624X) showing no LOH. Arrows show variants of interest on sequencing tracks