
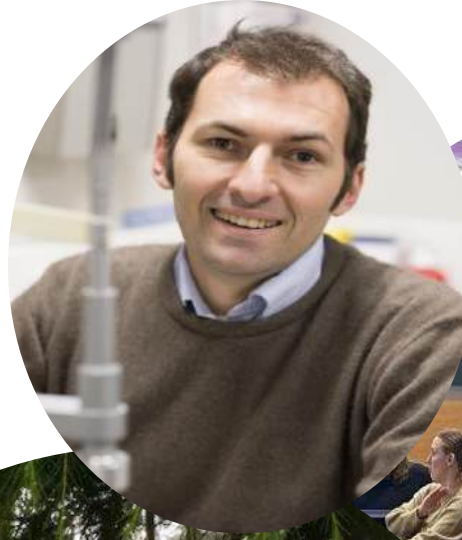




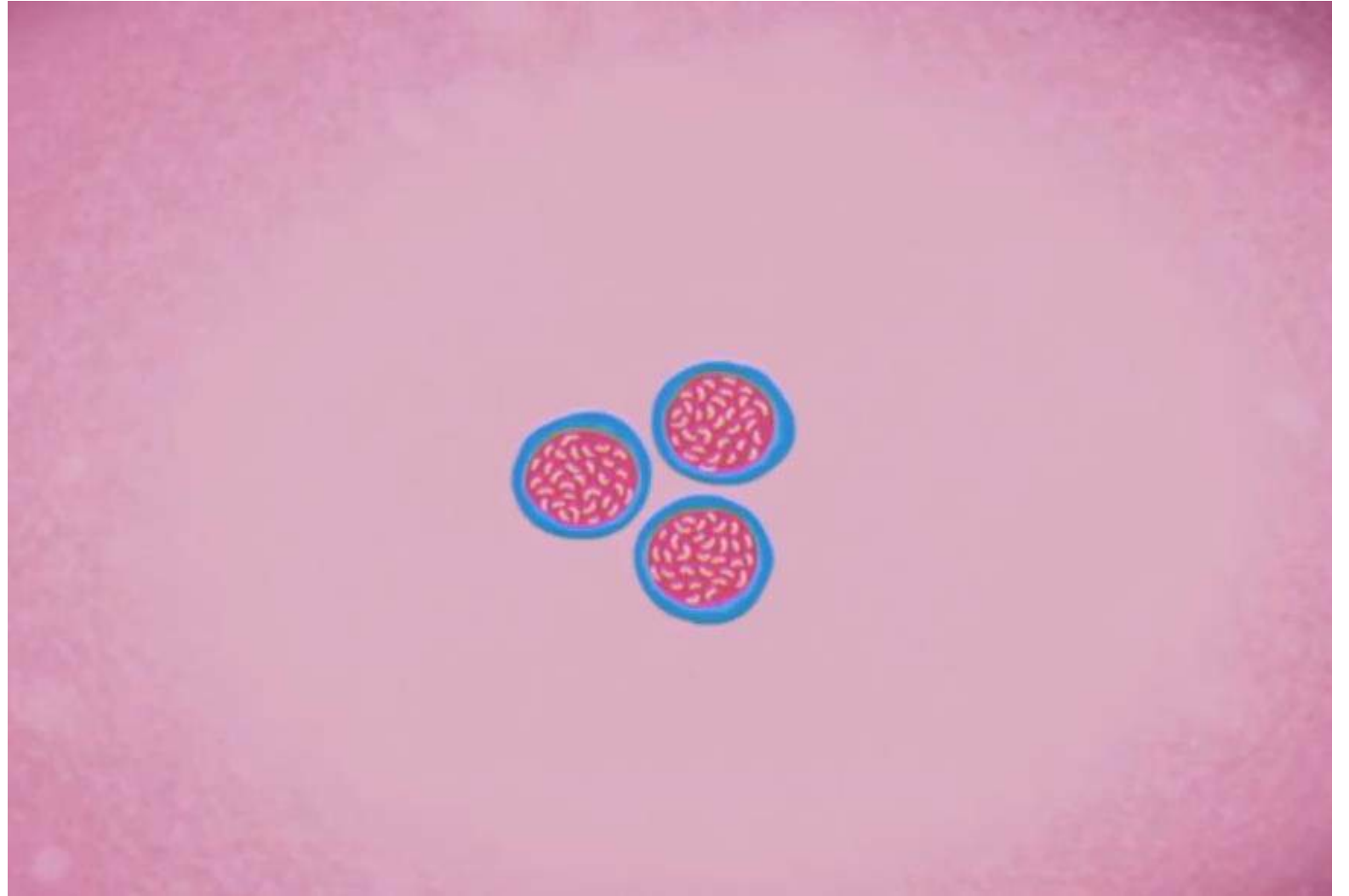
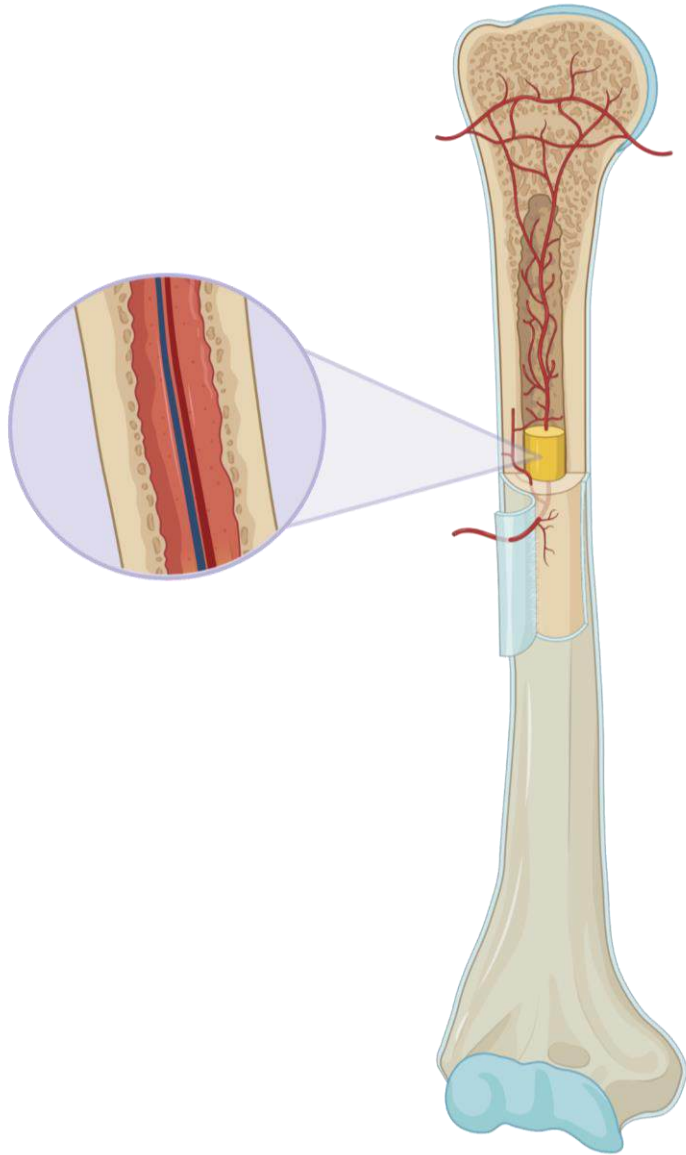
Bone Marrow Failure Research



Kirsten Fairfax
School of Medicine, UTAS, Hobart
 @FairfaxKirsten



UNIVERSITY of
TASMANIA



Smith M, 2020, *How bones make blood*, TED-Ed

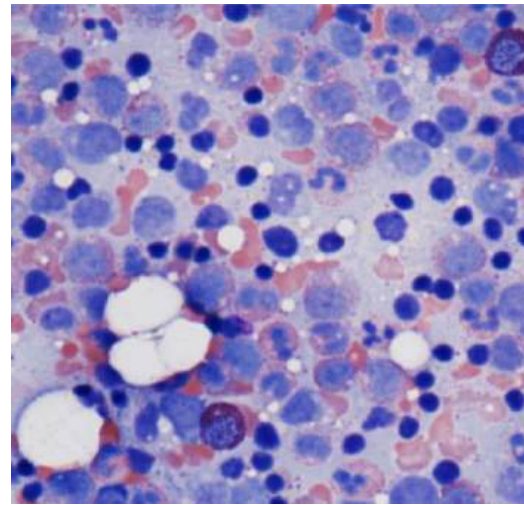
Inherited and Acquired BMFS

Inherited

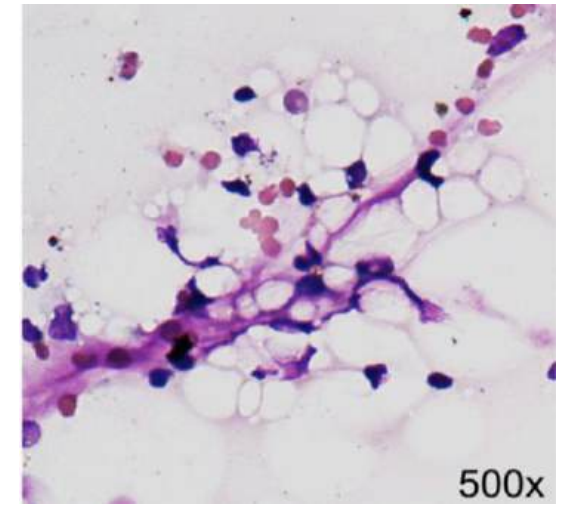
- Defects in DNA repair, telomerase and ribosomal pathways
- Can feature extra marrow presentations

Acquired

- Immunological attack on body's haematopoietic stem cells
- Thought to be triggered by environmental exposures



Normal bone marrow

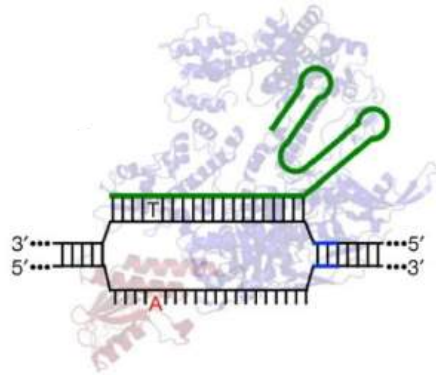


AA bone marrow

Wilson D, *et al.* 2014

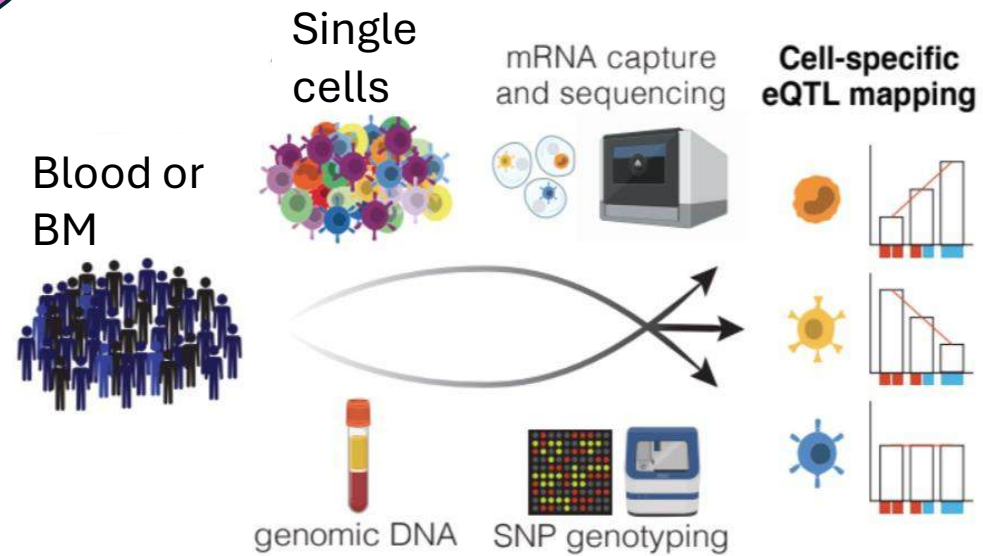
Sharma R, *et al.* 2016

CRISPR editing

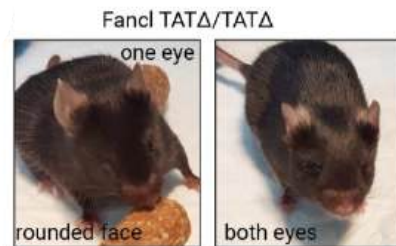
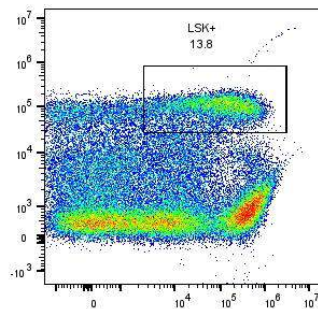


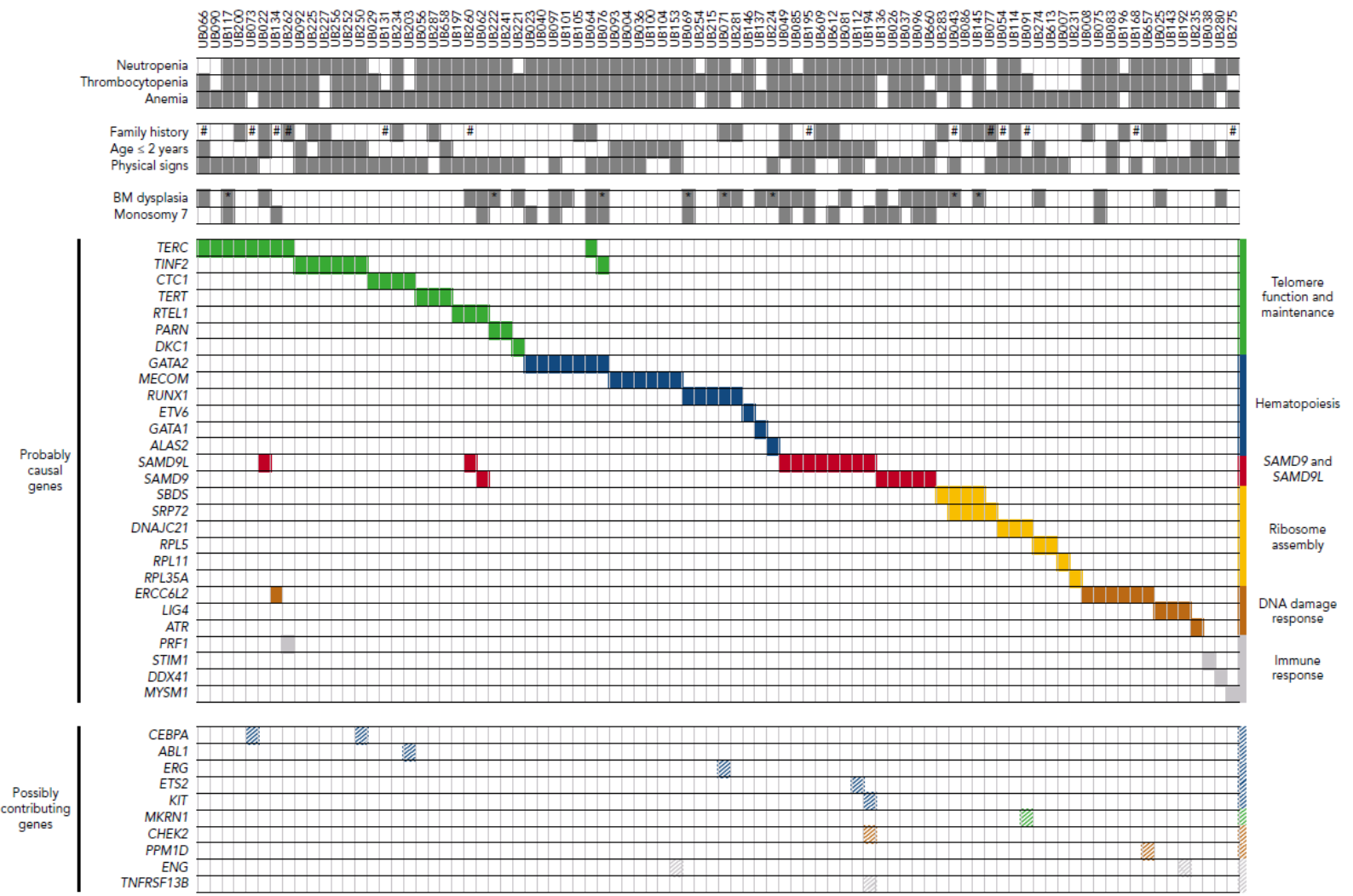
BMF research

Single cell eQTL mapping



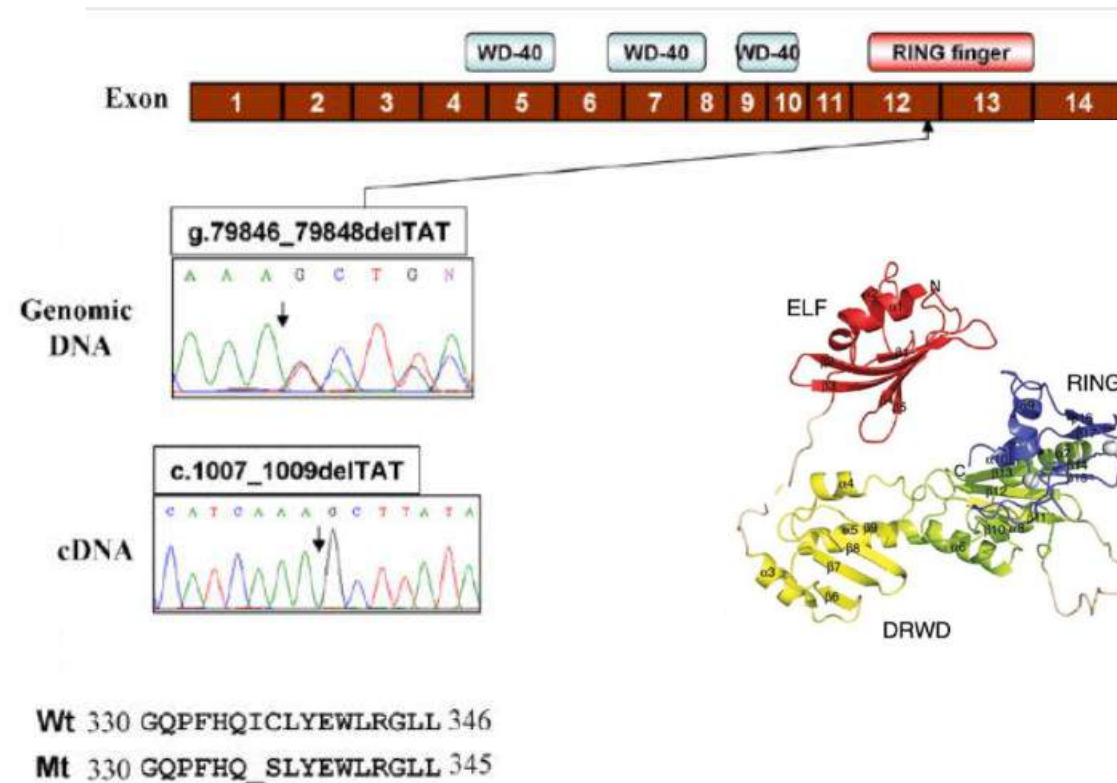
Mouse model analysis



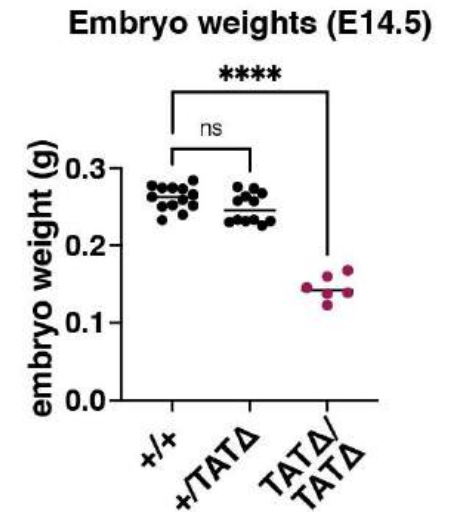
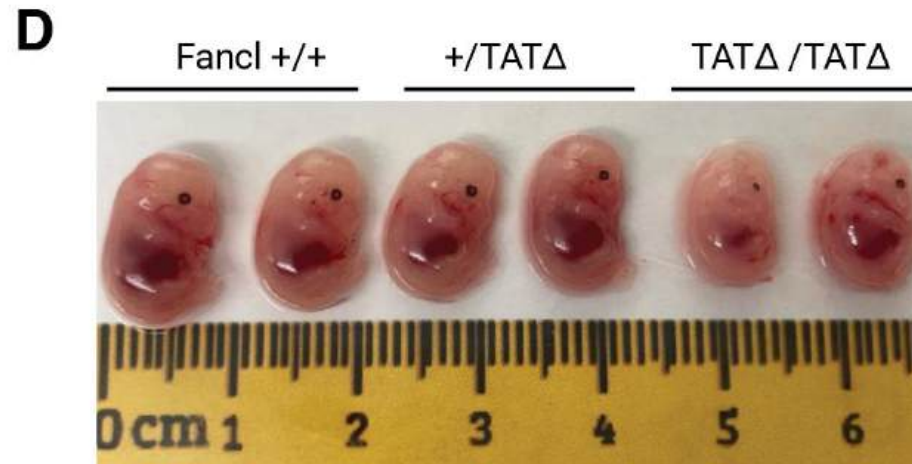
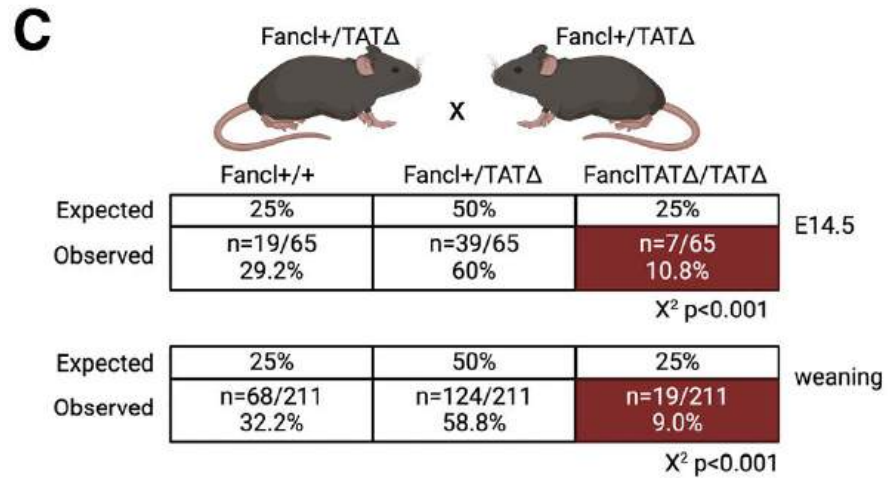
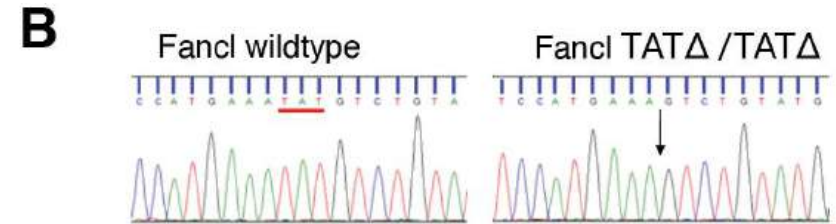
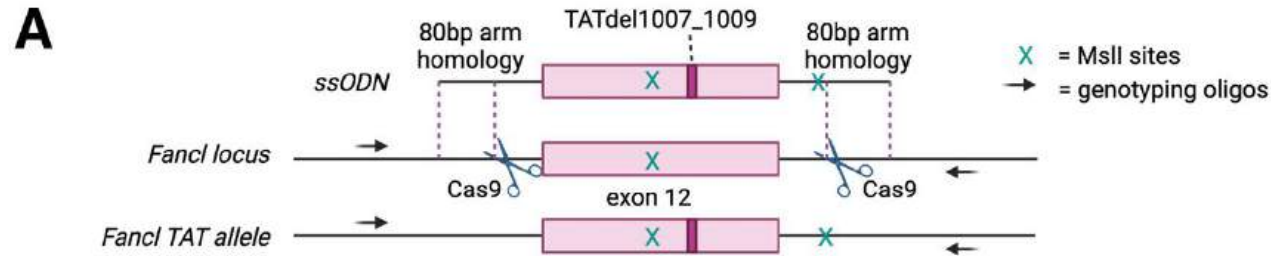


Bluteau et al., 2018

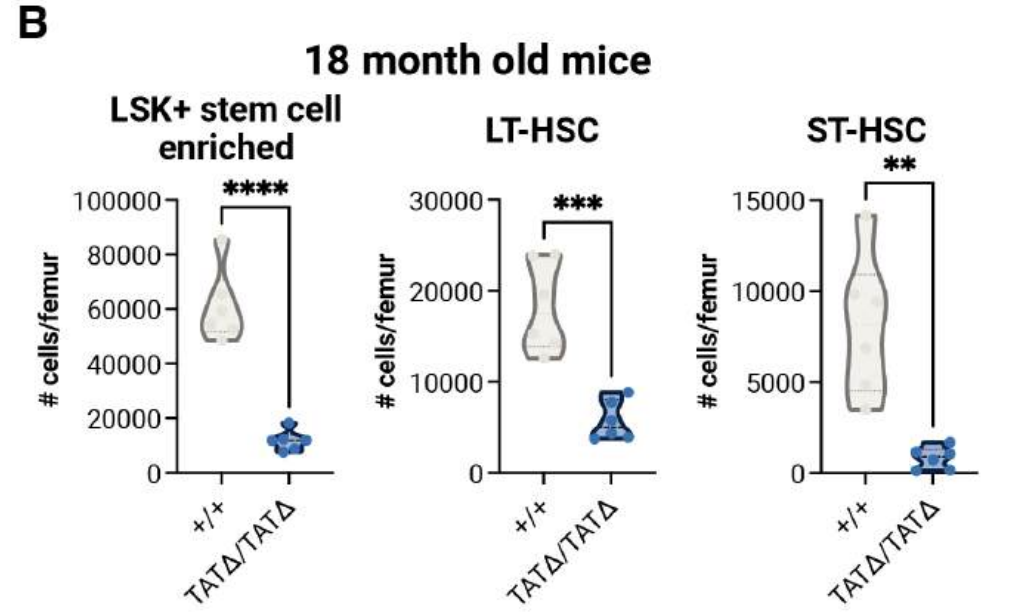
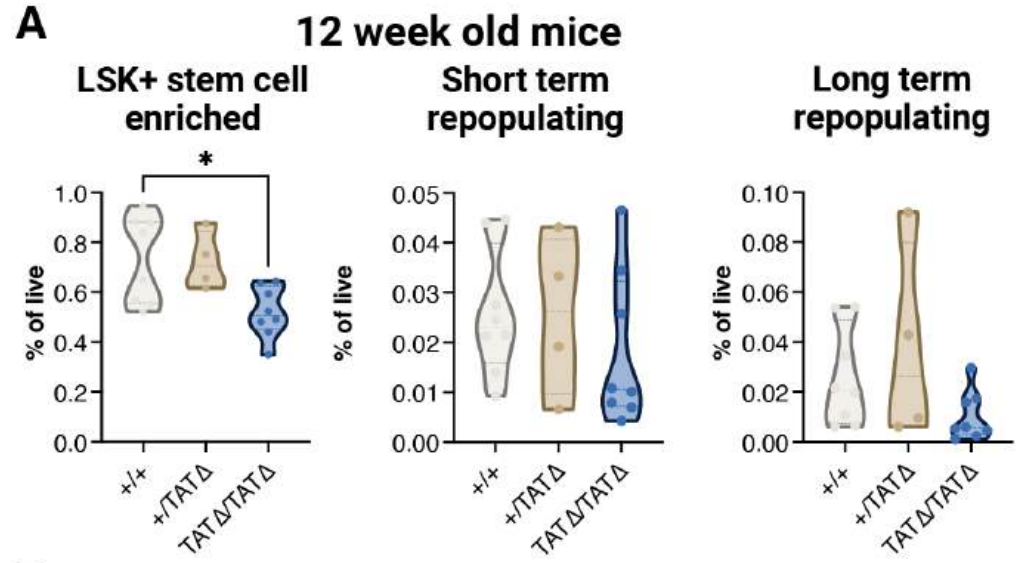
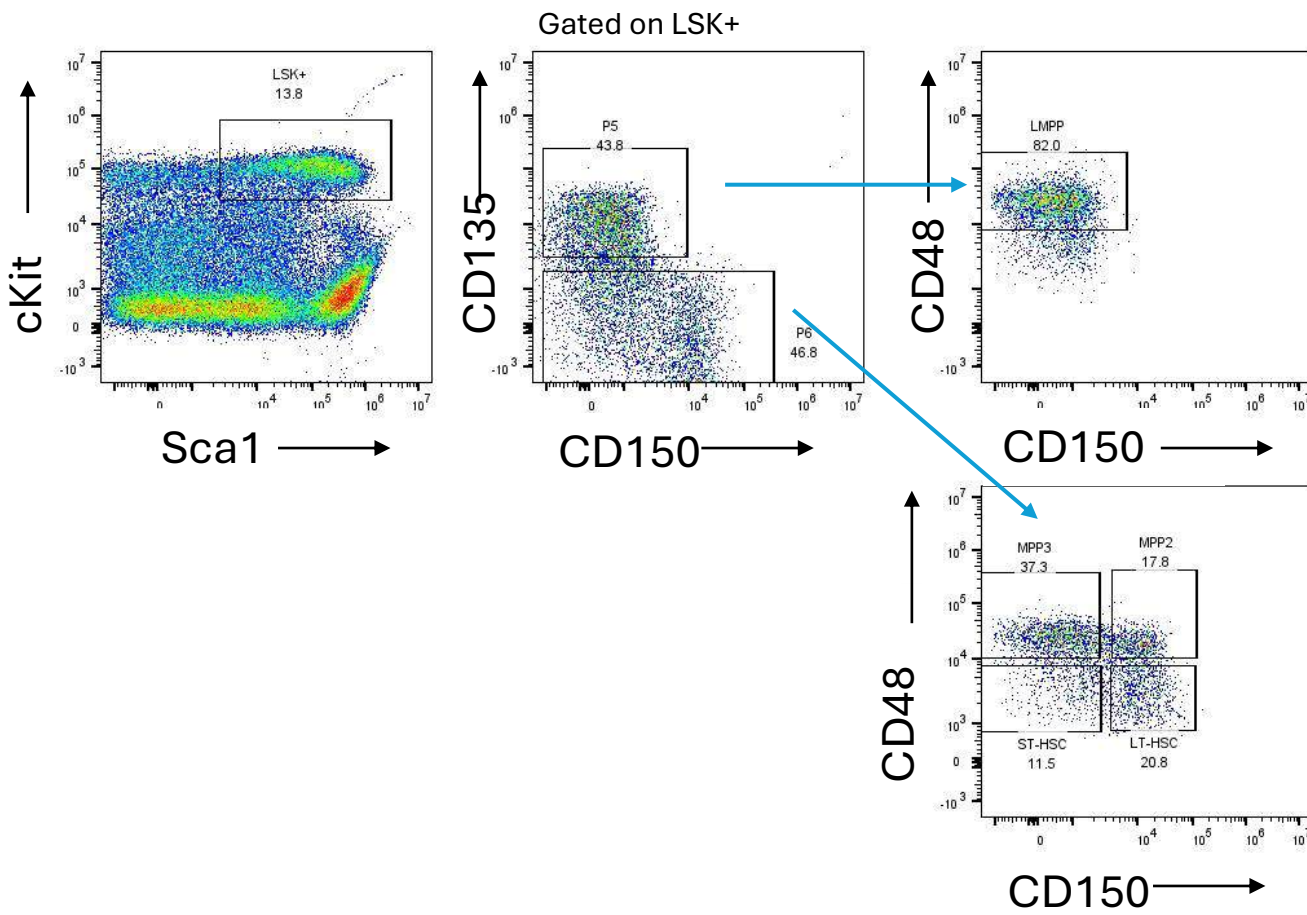
FancL-TAT deletion



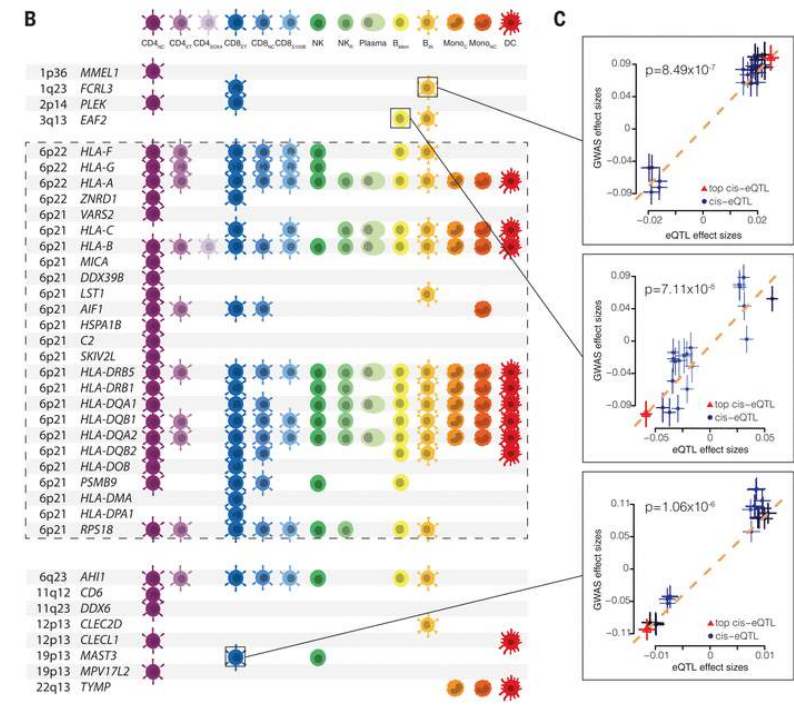
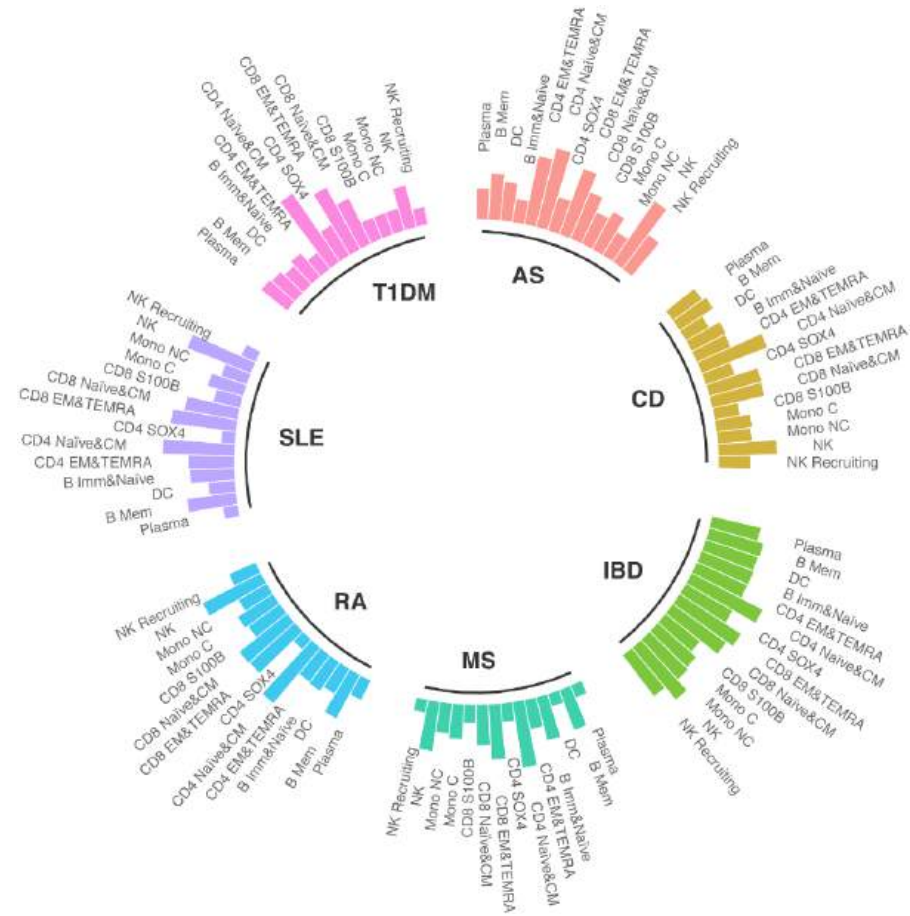
FancL^{TATΔ} mice have embryonic defects



FancL^{tatΔ} mice have stem cell defects

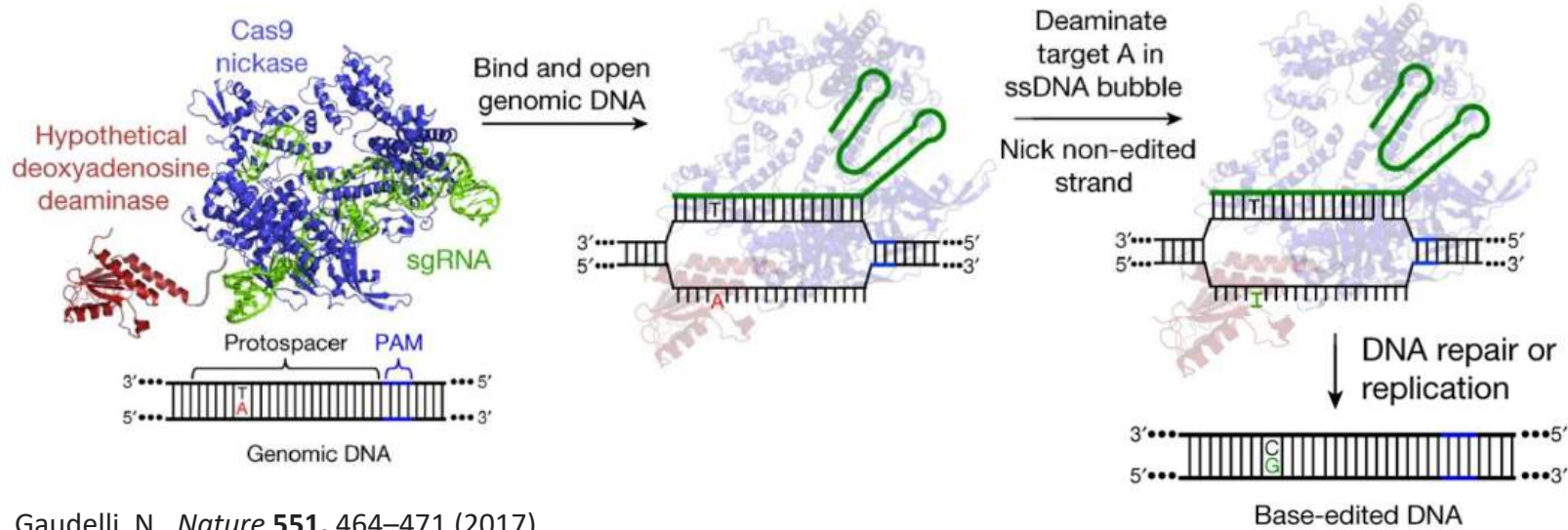
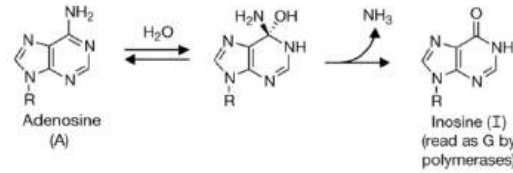


eQTL and Autoimmunity

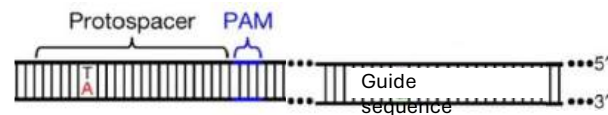


Seyhan Yazar et al. Science, 2023

Base editing

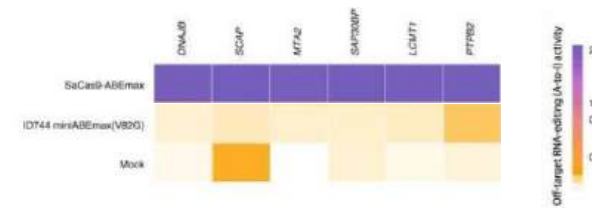


Gaudelli, N., *Nature* 551, 464–471 (2017).



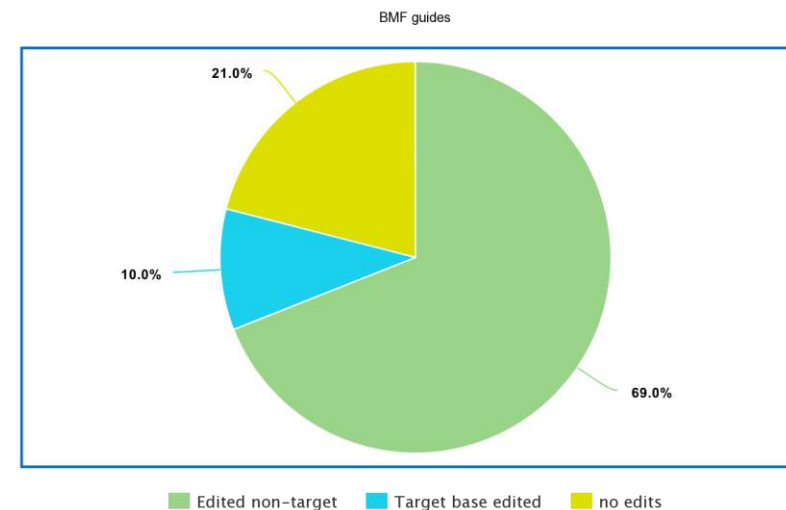
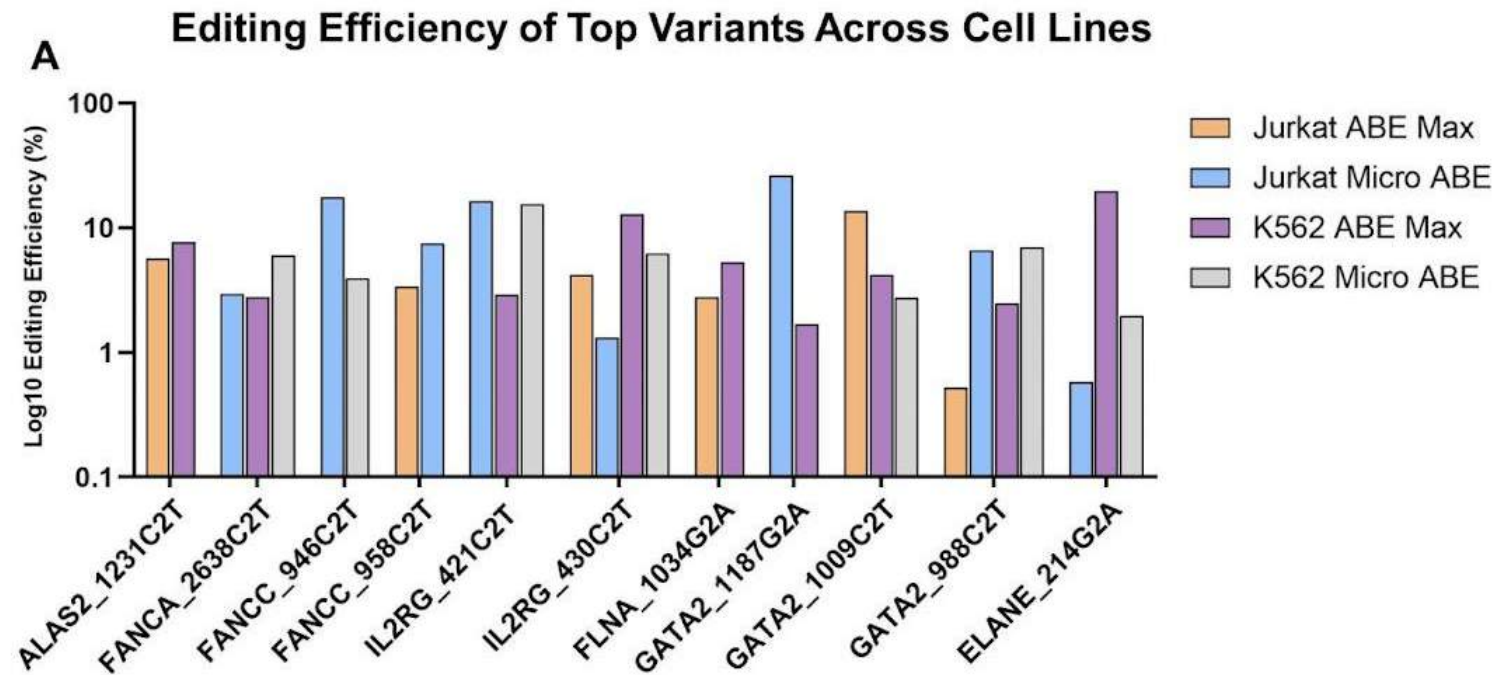
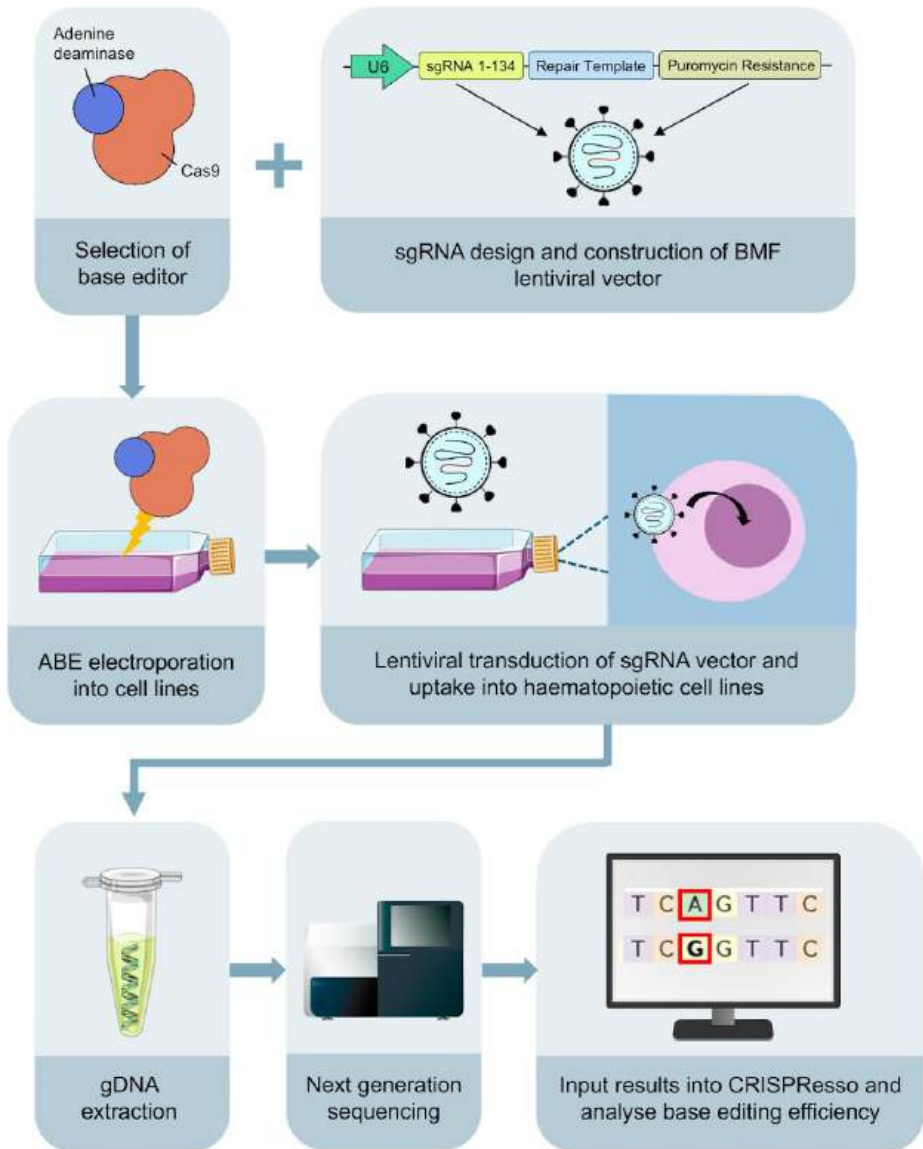
Library: iBMF sequenceguide targeting
iBMF sequence
FANCA:c.1027C>T (p.Gln343Ter)

Examples: RPL5: c.67C>T (p.Arg23Ter)
MPL: c.769C>T (p.Arg257Cys)



Tran, M., *Nature Commun* 11(1):4871 (2020).





Edited non-target Target base edited no edits

meta-chart.com

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participants!

