Long-Term Persistence of Donor-Derived Del(20q) : A Case Report and Literature Review

Clara Bouley[1], Min Fang [1,2], Kate Kroeger [2] , Jerald Radich[1,2], Judy Campbell [2], Paul A. Carpenter [1,2], Stephanie J. Lee [1,2], Marco Mielcarek [1,2], Rainer Storb [1,2] , Masumi Ueda Oshima[1,2]
[1,]University of Washington School of Medicine, [2]Fred Hutch Cancer Center

BACKGROUND

Deletion of the long arm of chromosome 20 (del(20q)) is a cytogenetic abnormality with potential for malignant transformation and is the most prevalent large structural genetic abnormality in older adults[1]. Although rare, del(20q) can arise in donor-derived cells after hematopoietic cell transplantation (HCT), but its occurrence in these cells is not well documented, with many cases lacking long-term follow-up. This study examines the persistence and clinical implications of donor-derived del(20q) to inform post-transplant monitoring. We hypothesize that while not all cases of donor derived del(20q) are benign, the majority will follow a benign course.

METHODS

Case Report: A woman received a hematopoietic stem cell transplant for angioimmunoblastic T-cell lymphoma in 1997. Early post-transplant, her donor was diagnosed with myelodysplastic syndrome associated with del(20q), and the recipient developed del(20q). Despite this, the recipient remains without overt myeloid neoplasm, now 26 years post-transplant. We present data from cell-fractionated FISH studies in the recipient at 20- and 26-years post-transplant, and next-generation sequencing of donor marrow (1997) and recipient blood (2018, 2024) using a myeloid neoplasm assay.

Literature Review: PubMed and Google Scholar were searched using terms related to del(20q), stem cell transplantation, and donor-derived cells. We performed complete text review of 18 PubMed citations and 40 Google Scholar citations from their respective query results. Inclusion criteria included confirmation of complete donor chimerism and del(20q) positivity above threshold (≥2/20 metaphases or ≥4% FISH signal). This is the largest collection of these cases to date.

RESULTS

Case Report

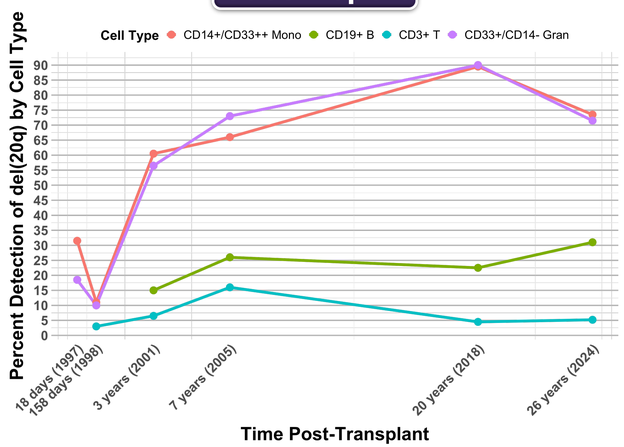
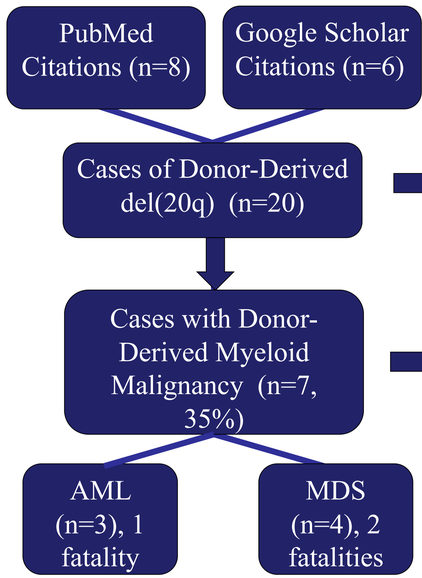


Fig. 1: Percent positivity of del(20q) in recipient cell fractions as detected by FISH.

Literature Review



Donor-Derived del(20q) Recipients (n=20)	Median	Range
Age of Recipient (years)	57	19-65
Time to Detection of del(20q) Post-transplant (months)	16	0.6-89
Follow-up Duration Post-transplant (months)	36	9-319

Recipients who developed Myeloid Malignancy (n=7)	Median	Range
Age of Recipient (years)	58	36-65
Time to Detection of del(20q) Post-transplant (months)	22	1.8-89
Time to Myeloid Malignancy Post-transplant (months)	22	7-89
Follow-up Duration Post-Transplant (months)	29	9-142

DISCUSSION

- We present the longest follow-up to date for a donor derived del(20q) case, showing that even with long-term high-level del(20q) positivity in myeloid cells it can remain benign.
- Minority of del(20q) cases progressed to myeloid malignancy.
  - Recipients with myeloid malignancies had a shorter follow-up duration, partly due to fatalities.
- The limited number of cases and incomplete follow-up in the literature constrains our ability to fully analyze del(20q) behavior in the post-transplant setting.

CONCLUSIONS

Del(20q) largely presents as a clonal cytogenetic abnormality of undetermined significance in the post-transplant setting. While our case demonstrated a benign outcome even with long-term positivity, the literature shows that malignancy can develop soon after del(20q) detection. Regular follow-up, particularly in the early months post-detection, is crucial to assess the risk of progression to malignancy.

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REFERENCES

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Plain Language Summary

Our study focuses on a patient who received a bone marrow transplant and developed a chromosomal change, del(20q), from the donor's cells. Despite this change, the patient has not developed any related types of cancer. We reviewed similar cases and found that while most remain healthy, there is still a risk of developing cancer, so careful monitoring is important.