

KAT6 Biobank Status

KAT6 patient specific iPSC Bank | Founded in 2022

Serrano Lab | CReM | KAT6 Foundation

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1 Background

1.1 The Importance of Patient-Specific iPSC Banks for Rare Disorders

Induced pluripotent stem cells (iPSCs) are a powerful tool in biomedical research, allowing scientists to derive stem cells from somatic (non-stem) cells of individual patients. iPSCs can be differentiated into various cell types, enabling disease modeling, drug screening, and personalized therapeutic strategies. When it comes to **rare disorders**, such as those associated with KAT6A and KAT6B pathogenic variants, patient-specific iPSC lines are particularly valuable. They capture the exact genetic makeup of individuals, allowing a more precise understanding of disease mechanisms and a platform for testing potential treatments tailored to each patient’s molecular background.

1.2 Open Source Biology at the CReM and Serrano Lab

The [Center for Regenerative Medicine \(CReM\)](#) and [The Serrano Lab](#) at Boston University embraces a philosophy of **open source biology**, meaning that research findings, protocols, and biological resources (such as iPSC lines) are shared openly with the broader scientific community to accelerate discoveries and therapy development. By making these resources freely available, the CReM and the Serrano Lab foster collaboration, reduce duplication of effort, and speed the translation of basic research into clinical applications. This approach ensures that patients, researchers, and the global community benefit from scientific progress as quickly and equitably as possible.

1.3 Patient-Partnered Research: The KAT6 Foundation Collaboration

The collaboration between the [KAT6 Foundation](#), the Serrano Lab, and the CReM is a prime example of **patient-partnered research**. In this model, patient advocacy groups, researchers, and clinicians work together from the earliest stages of project design, thereby ensuring that the needs, priorities, and experiences of the patient community shape the scientific goals. This **iPSC biobank** of patient-derived samples of individuals with KAT6A and KAT6B variants, exemplifies that collaboration.

2 Status of KAT6 iPSC Bank

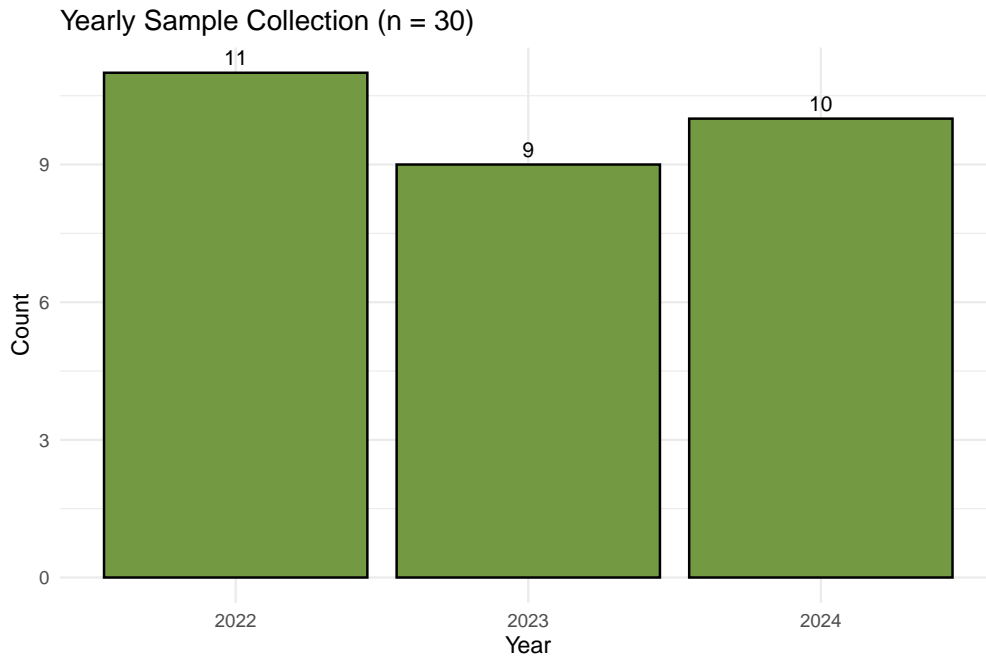
The outlined plots represent the status of the iPSC bank as per February 2025

Table 1: Participant Data

VariantID	AgeGroup	Sex	RacialEthnicIdentity	Gene	YearCollection	iPSC Line
A15	0-10	Male	White	KAT6A	2023	AWAITING
A14	11-20	Male	White	KAT6A	2023	UNSUCCESSFUL
A13	11-20	Female	Prefer not to say	KAT6A	2022	IN PROGRESS
A7	0-10	Male	Multiracial	KAT6A	2022	AWAITING
A9	0-10	Male	White	KAT6A	2022	AWAITING
A3	11-20	Male	Black or African American	KAT6A	2023	YES
A1	0-10	Male	White	KAT6A	2023	AWAITING
A10	11-20	Male	Prefer not to say	KAT6A	2022	YES
A6	0-10	Male	White	KAT6A	2022	AWAITING
A12	0-10	Male	Multiracial	KAT6A	2022	IN PROGRESS
A2	0-10	Male	White	KAT6A	2023	AWAITING
A11	11-20	Male	White	KAT6A	2022	AWAITING
A8	0-10	Female	Multiracial	KAT6A	2022	YES
A5	0-10	Female	White	KAT6A	2022	AWAITING
A4	11-20	Female	White	KAT6A	2022	UNSUCCESSFUL
A16	31-40	Female	White	KAT6A	2024	AWAITING
A17	31-40	Male	White	KAT6A	2024	AWAITING
A18	0-10	Male	White	KAT6A	2024	AWAITING
B1	11-20	Male	White	KAT6B	2023	UNSUCCESSFUL
B2	0-10	Female	Prefer not to say	KAT6B	2023	YES
B3	11-20	Male	White	KAT6B	2022	YES
B4	0-10	Female	White	KAT6B	2023	AWAITING
B5	0-10	Male	White	KAT6B	2023	YES
B6	0-10	Male	White	KAT6B	2024	AWAITING
B7	0-10	Female	White	KAT6B	2024	AWAITING
B8	21-30	Female	White	KAT6B	2024	IN PROGRESS
B9	11-20	Male	Hispanic or Latino	KAT6B	2024	AWAITING
B10	21-30	Female	Prefer not to say	KAT6B	2024	AWAITING
B11	0-10	Female	White	KAT6B	2024	IN PROGRESS
B12	0-10	Female	White	KAT6B	2024	AWAITING

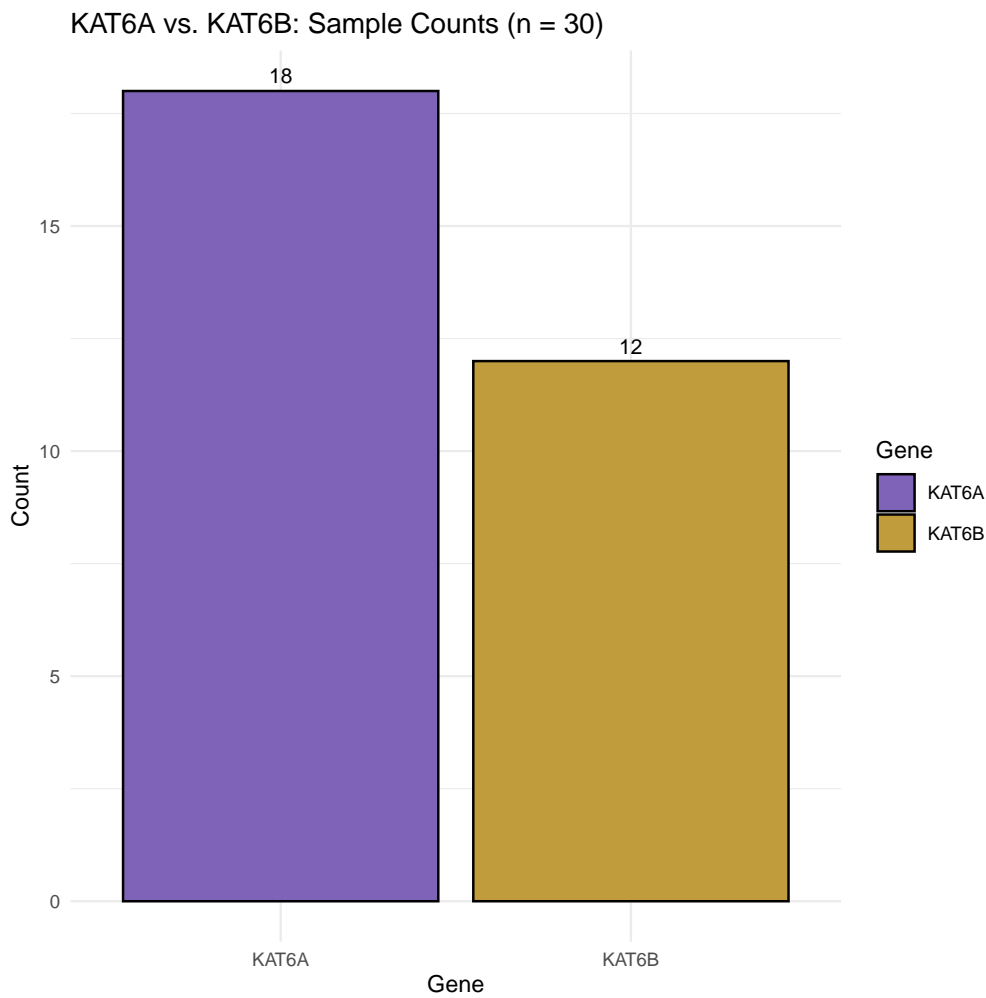
2.1 1. Samples Collected by Year

This bar plot compares the yearly sample collection. Counts are displayed above each bar.



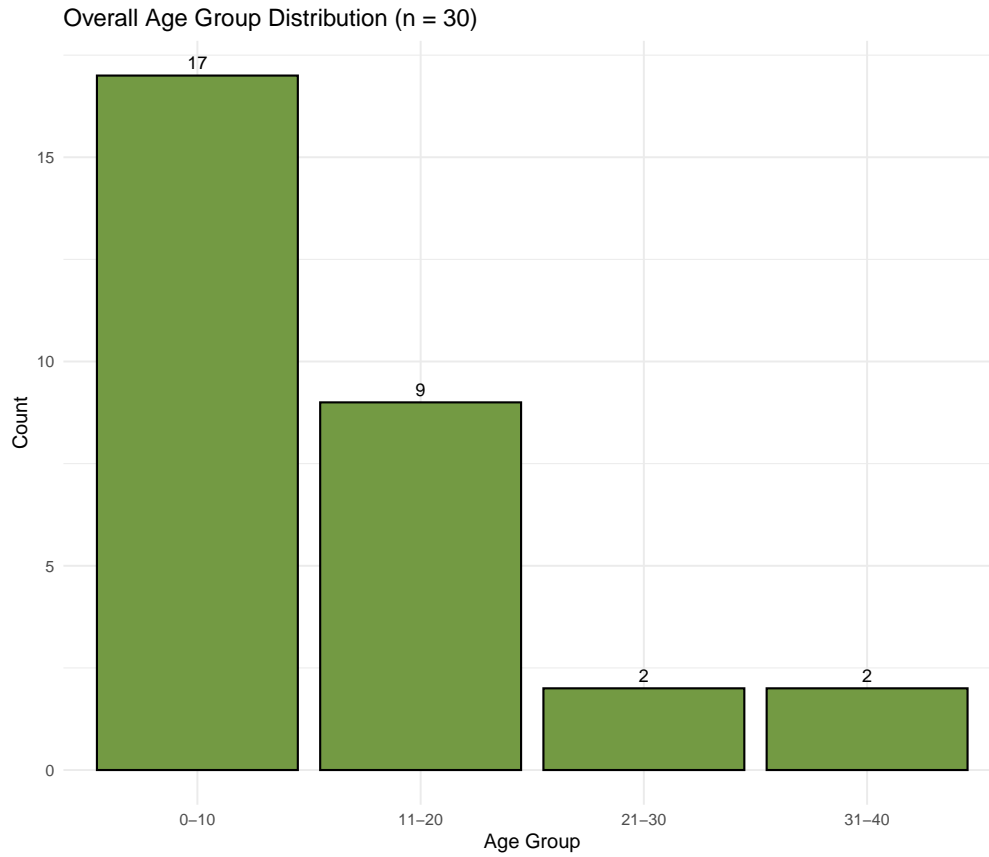
2.2 2. Comparison of Gene Counts (KAT6A vs. KAT6B)

This plot compares the number of samples for **KAT6A** versus **KAT6B**.

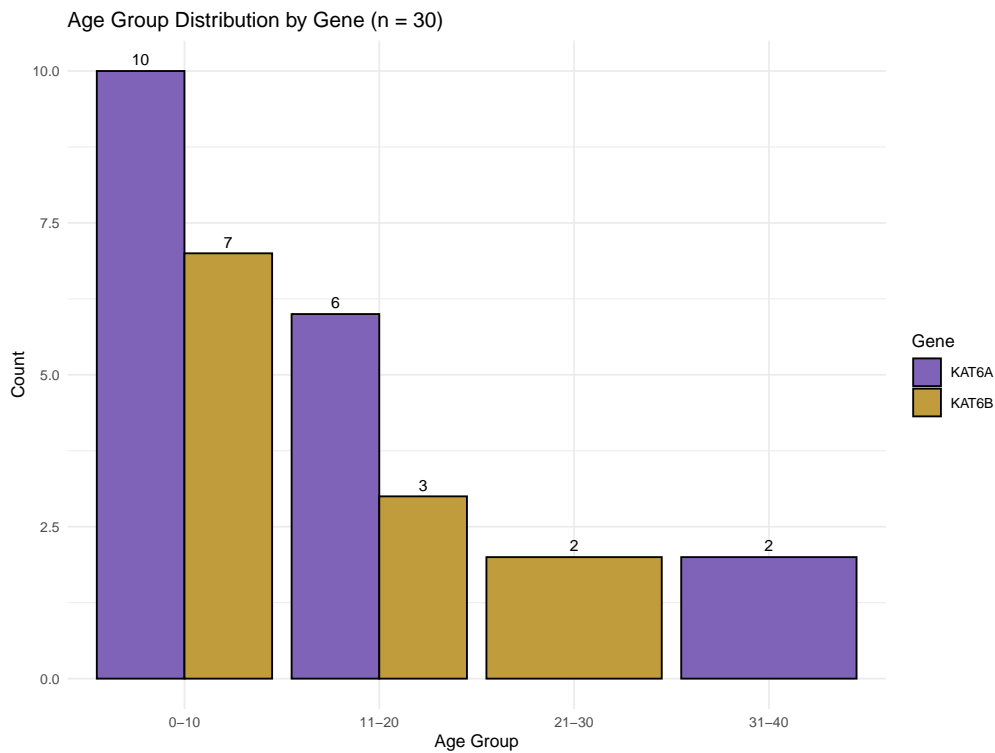


2.3 3. Age Group Distribution

2.3.1 (a) Overall Age Group Distribution

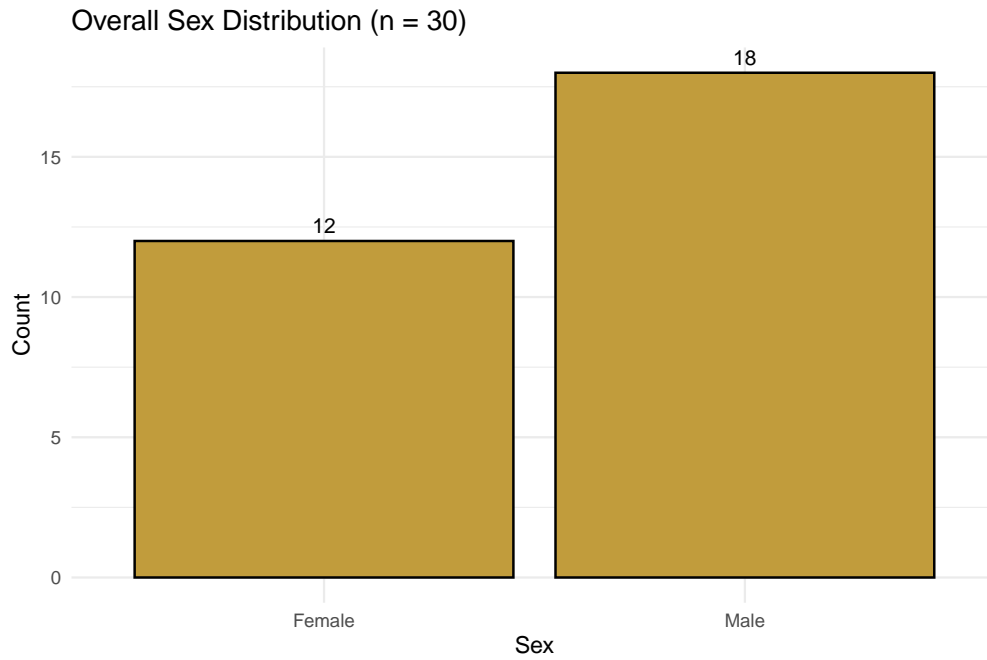


2.3.2 (b) Age Group Distribution by Gene

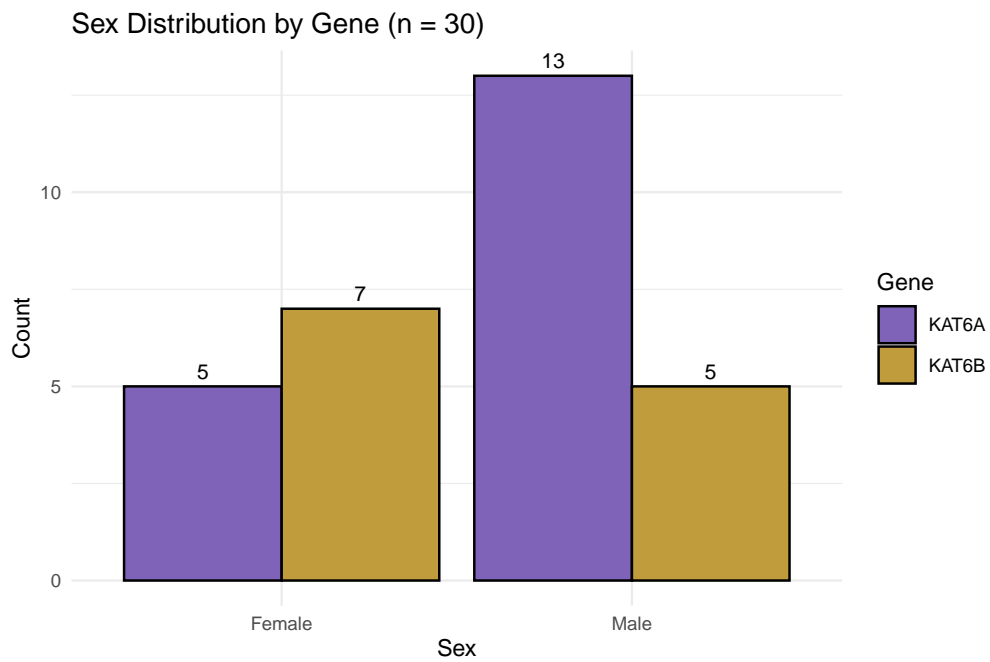


2.4 4. Sex Distribution

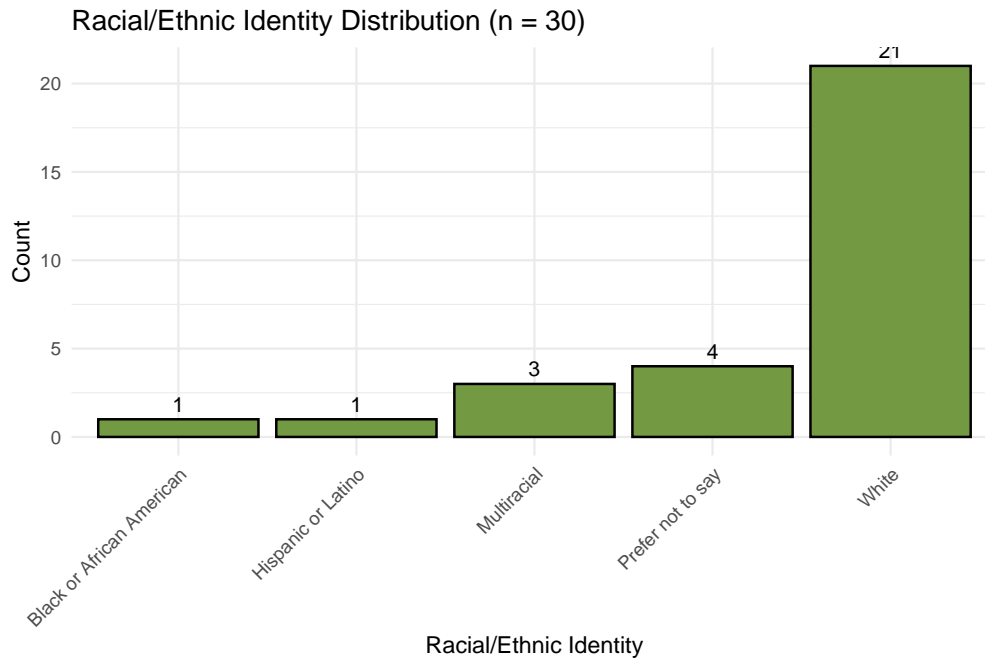
2.4.1 (a) Overall Sex Distribution



2.4.2 (b) Sex Distribution by Gene

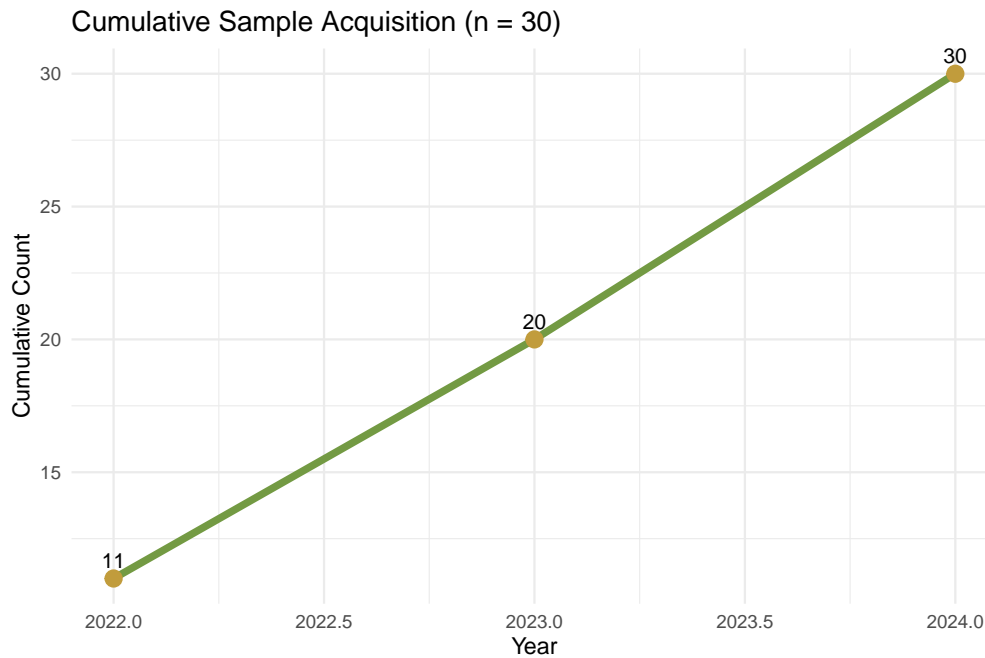


2.5 5. Racial/Ethnic Identity Distribution

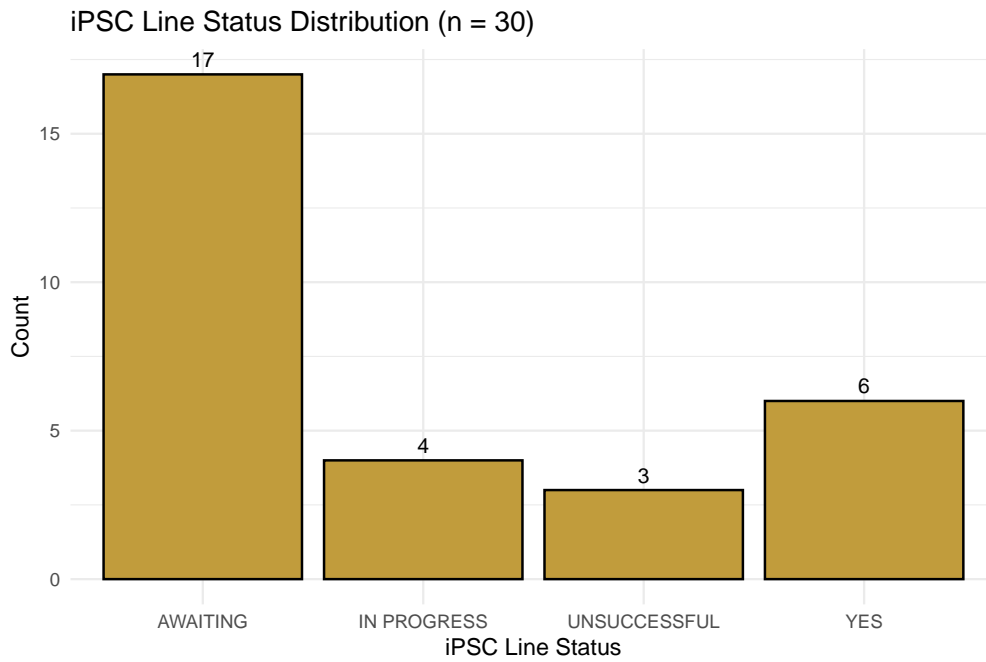


2.6 6. Cumulative Progression of Samples Acquired

This line plot shows the cumulative sample count over the years.



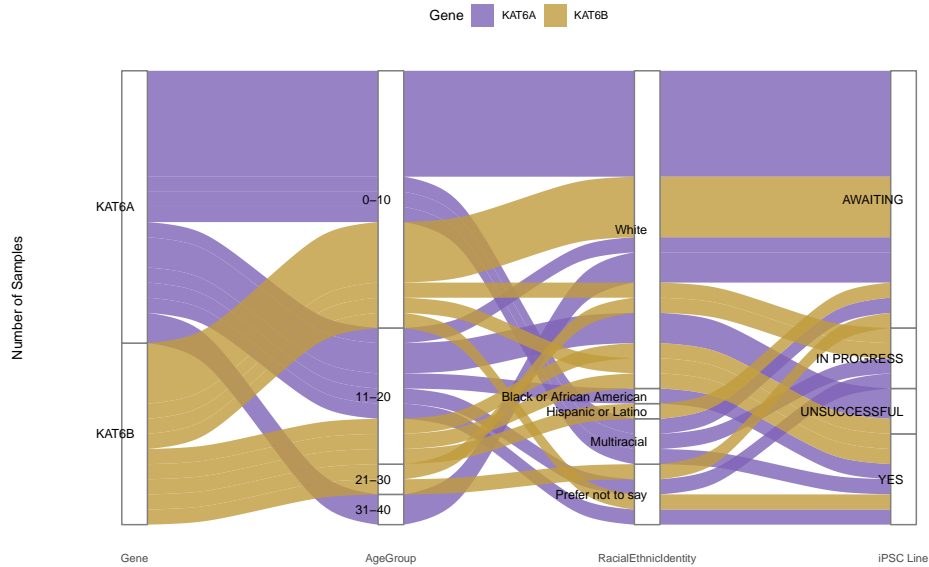
2.7 7. iPSC Line Status Distribution



2.8 8. Overview Diagram

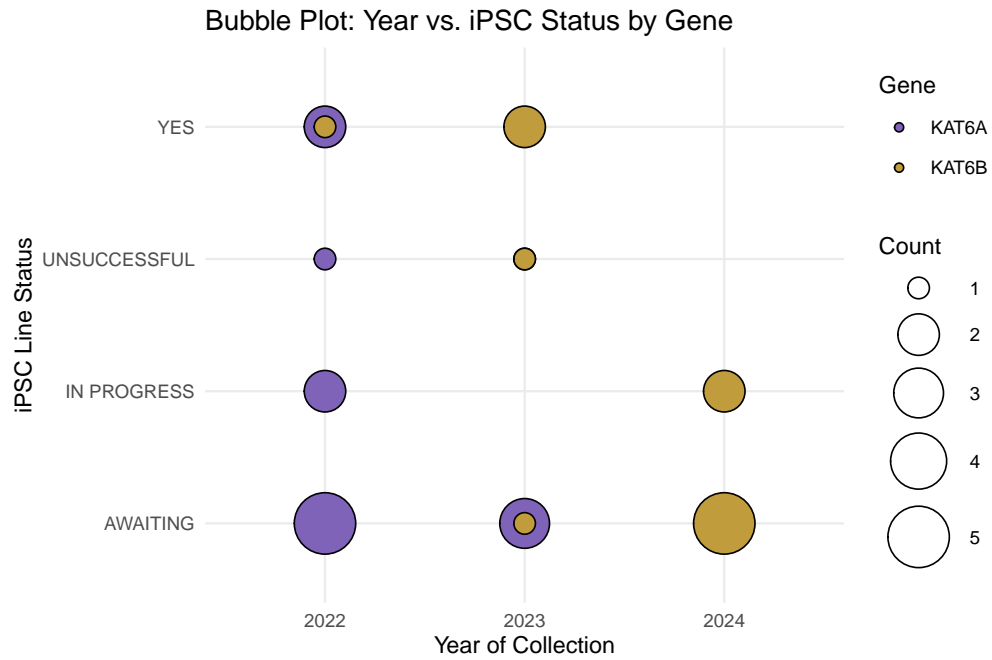
This alluvial diagram shows how samples flow from **Gene** to **Age Group**, then to **Racial/Ethnic Identity**, and finally to **iPSC Status**. The width of the flows is proportional to the sample count.

Alluvial Diagram: Gene, Age, Ethnicity, & iPSC Status (n = 30)



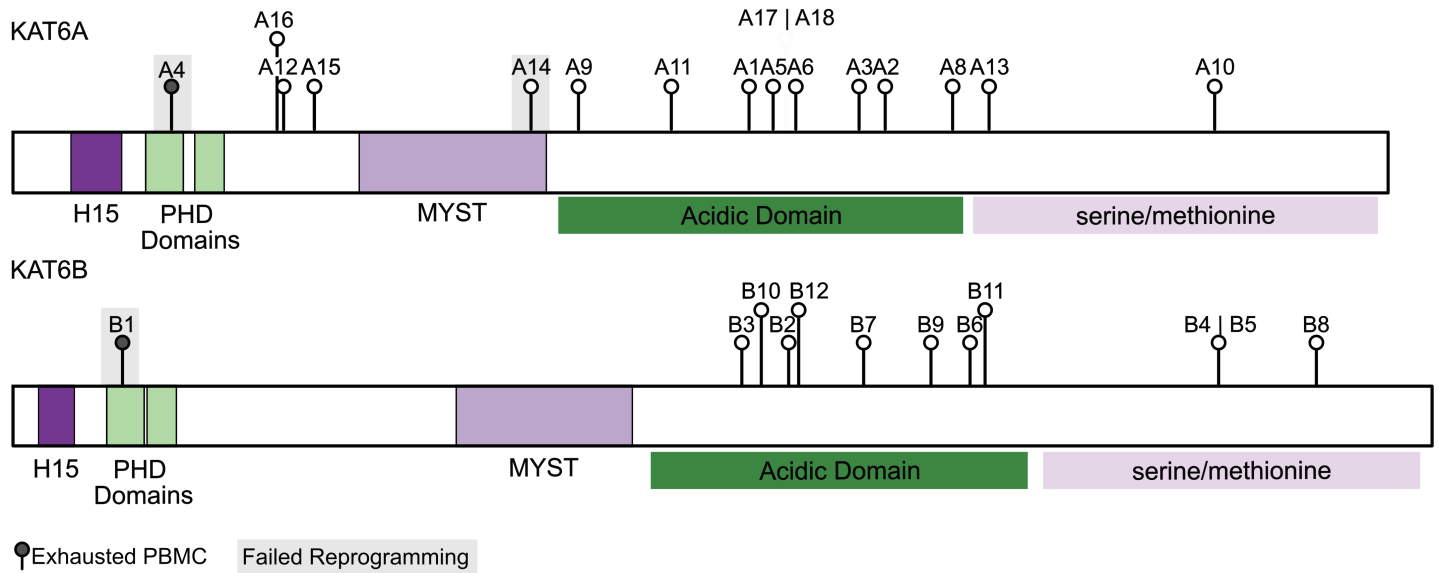
2.9 9. Overview of iPSC line Reprogramming Status Each Year

This bubble plot shows the number of samples by **YearCollection** and **iPSC Line Status**, with bubbles colored by **Gene**.

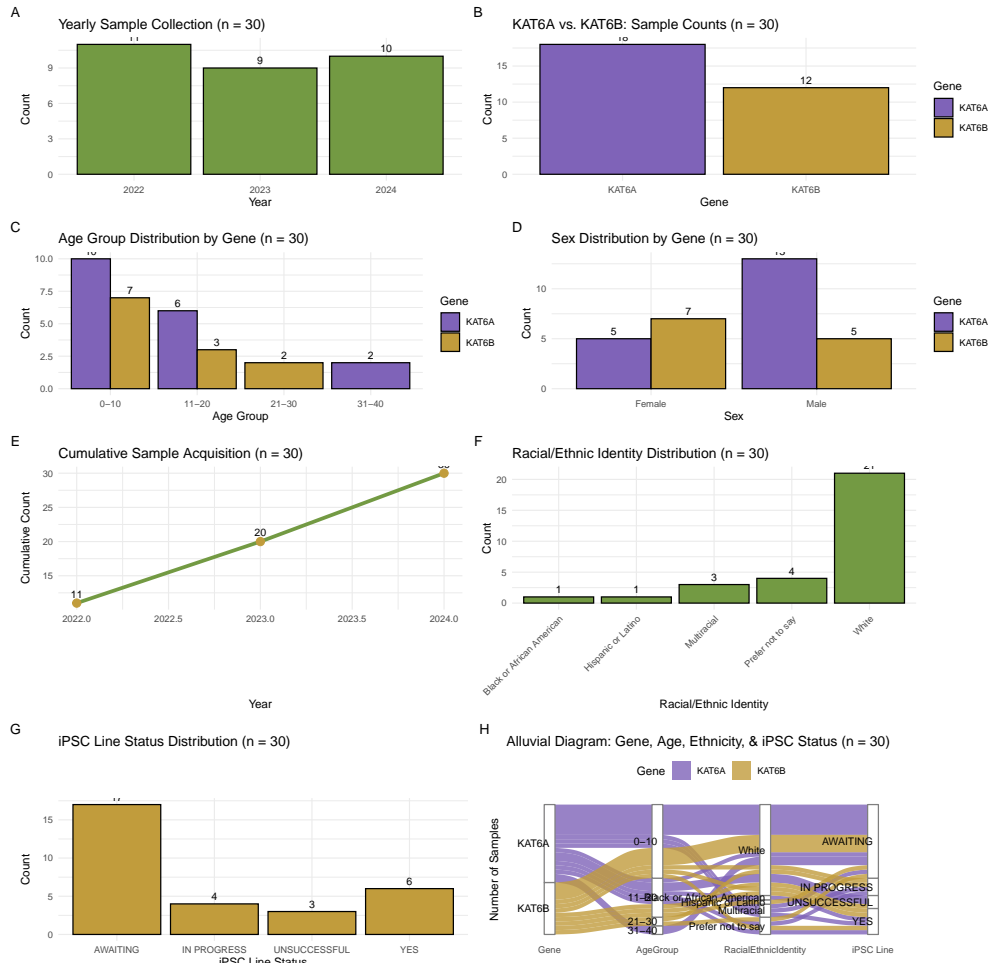


3 Summary

KAT6A and KAT6B Variants Present in the Bank



Composite Summary of KAT6 Biobank Data



4 Support the KAT6 iPSC Bank

If you are a member of the KAT6 community and would like to learn more about this project or participate in the KAT6 iPSC bank, we will be recruiting participants at the upcoming [6th KAT6 International Conference](#). To pre-register or to indicate your interest, please [complete this questionnaire](#).

Make a monetary donation to support our cell sharing mission.

If you would like to help grow the KAT6 iPSC bank financially, please consider a donation through the link below. When prompted, kindly add **KAT6/Serrano Lab/CRem** in the note field so we can direct your gift toward this initiative:

[DONATE](#)