

TRACING THE DEVELOPMENTAL ORIGINS OF ATYPICAL TERATOID Rhabdoid TUMOR TYR SUBTYPE

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2. STUDY DESIGN

-Only one ATRT-TYR [1] snRNA-seq available

1. BACKGROUND

- Atypical teratoid rhabdoid tumor (ATRT) is a rare and aggressive pediatric brain tumor
- Characterized by SMARCB1 (95%) or SMARCA4 (5%) mutations
- ATRTs represent 3% of the CNS pediatric tumors & 20% of patients under 3 years old.
- They are comprised of 3 distinct molecular subtypes: SHH, MYC and TYR.
- 70% of cases arise in the infratentorial region of the brain
- Very little is known about their developmental origins (cell of origin and the time of onset)
- TYR subtype is understudied and lacks murine models

SubType	FREQUENCY	SEX	AGE	LOCALIZATION	SMARCB1 MUTATION	SIGNATURES GENES
SHH	~ 40 %	M - 55 % F - 45 %	Median: 20mth	65% 35%	Point mutations/ Focal deletions	Neurogenesis SHH signaling NOTCH signaling
MYC	~ 25 %	M - 55 % F - 45 %	Median: 27mth	50% 38%	Broad deletions	MYC, HOX cluster genes
TYR	~ 35 %	M - 55 % F - 45 %	Median: 12mth	25% 75%	Point mutations/ Focal deletions	BMP signaling Melanogenesis (TYR, MITF) Mesenchymal genes (OTX2)

3. RESULTS

1 Tumor & micro-environment

ATRT heterogeneity

