



Impact of In Utero Transfusions in Fetuses with Hydrops Fetalis due to Alpha Thalassemia



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Background

- Alpha thalassemia major (ATM) is an inherited anemia that typically leads to hydrops fetalis.
- Emerging evidence suggests in utero transfusions (IUTs) can improve outcomes in this condition, which is often fatal in utero.

Objective

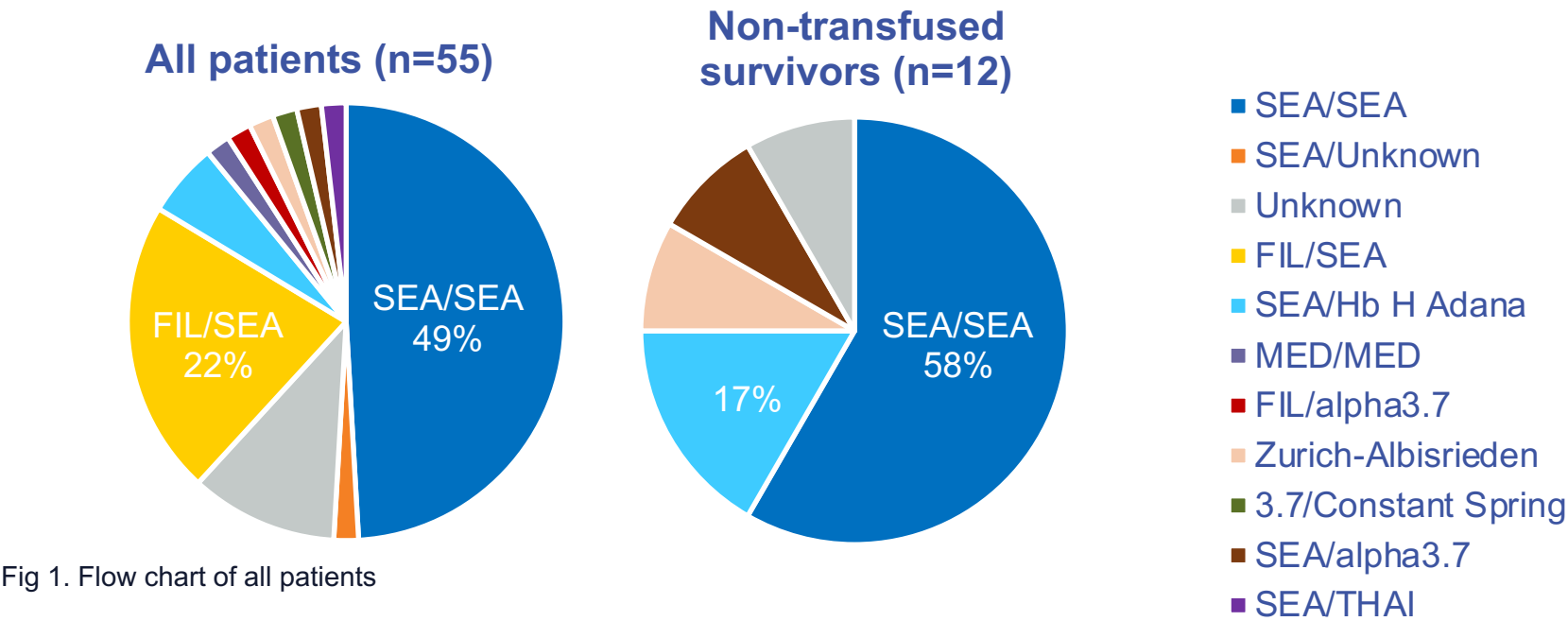
- To develop an international registry in order to understand the role of fetal therapy for alpha thalassemia major.

Study Design

- We compiled an international registry of alpha thalassemia variants from multiple tertiary care centers.
- Data included age at diagnosis, molecular diagnosis, number of IUTs, GA at delivery, and perinatal outcomes.
- Groups were compared using Mann-Whitney test.

Results

- Patients that underwent first transfusion at less than 24 weeks gestation had improved outcomes.
- All survivors with 4-gene mutations required serial transfusions, similar to those with beta thalassemia; 2 were cured by transplantation.
- All surviving neonates had low rates of congenital anomalies, composed primarily of mild cardiac defects and hypospadias.
- Of the transfused patients who were evaluated for neurocognitive development, all were normal.



- No patients who survived without intervention had the FIL deletion.

Performing IUTs in fetuses with alpha thalassemia major results in survival to near-term birth in almost all patients, with shorter hospital stays when compared to non-transfused.

***Two patients also underwent fetal hematopoietic stem cell transplantation.**

We are currently performing a phase 1 trial of in utero transplantation for ATM. For referrals: 1-800-RxFetus



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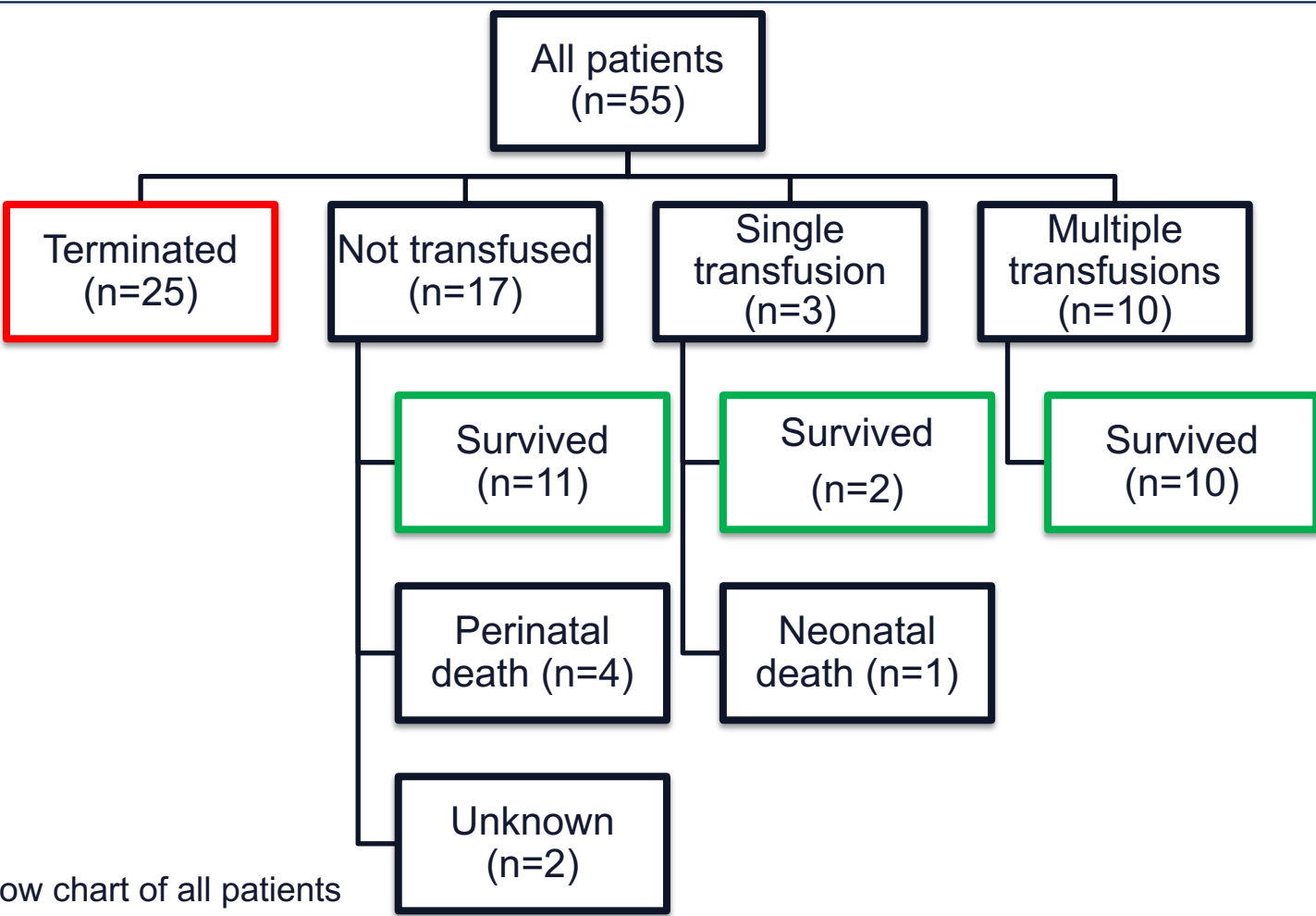


Fig 2. Flow chart of all patients

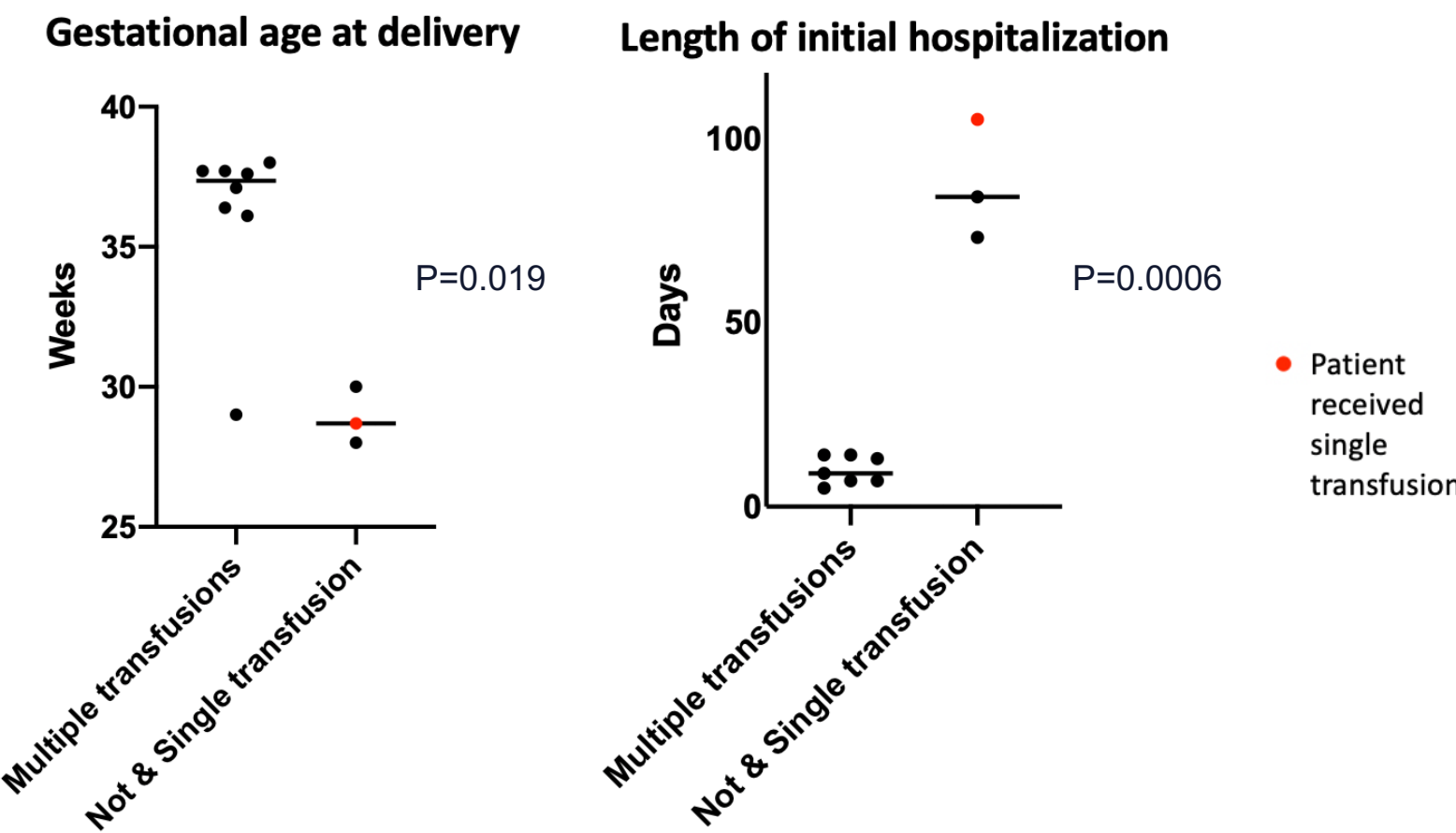


Fig 3. Multiple vs not/single transfusion survivors with 4-gene deletions

Patient ID	# IUTs total	GA of 1st IUT (weeks)	Pre-transfusion Hb (g/dL)	Transfusion volume (mL)	Standard transfusion volume (mL)	% of standard transfusion volume
Mean ± SEM	4.2 ± 0.5	23.5 ± 0.9	5.5 ± 0.5	26.4 ± 5.9	35.3 ± 4.4	66.7 ± 11.4

Fig 4. IUT characteristics