SAFETY AND EFFICACY OF FERTILITY PRESERVATION IN SICKLE CELL DISEASE AND TRANSFUSION DEPENDENT THALASSEMIA PATIENTS UNDERGOING GENE EDITING THERAPIES

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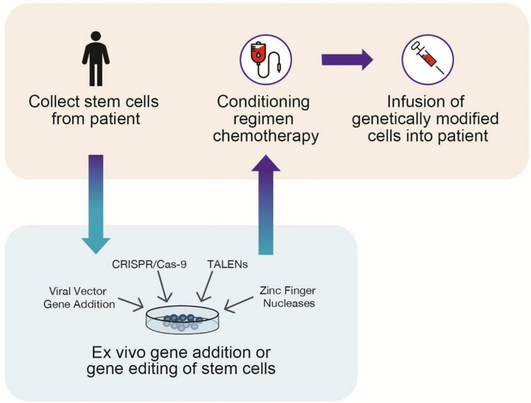
Background

- Sickle cell disease (SCD) and transfusion dependent β-thalassemia (TDT) are autosomal recessive disorders caused by genetic mutations that impair the function of the hemoglobin β subunit
- Advancements in gene therapy (GT) offer the potential for a functional cure of these conditions. However, the use of busulfan conditioning can result in infertility, making fertility preservation (FP) a critical component of multidisciplinary care
- Despite its importance, there is very limited data on the safety and efficacy of FP in individuals with SCD and TDT

Material & Methods

- Retrospective case series of patients with SCD and TDT undergoing FP prior to GT
- Participants in the trial were offered FP services via oocyte or sperm cryopreservation at Nashville Fertility Center
- Inclusion: pubertal patients without contraindication to oocyte or sperm cryopreservation
- Following FP consultation, individualized care plans were developed for each patient



Results

Table 1: Demographics & FP outcomes	SCD	TDT
Age (yrs)	24.2 (11-34)	18.9 (12-33)
Sex	8 females 6 males	9 females 8 males
Mean AMH (ng/mL)	2.6 (0.6-7.3)	2.8 (1.1-4.26)
Mean cycles ovarian stim	1.3 (1-2)	1.4 (1-3)
Mean oocytes retrieved	19 (6-42)	14 (3-30)
Mean oocytes cryopreserved	15 (6-37)	11 (3-24)
Mean duration gonadotropin treatment (days)	10 (7-14)	9.6 (6-14)
Mean total FSH dose (IU)	2311 (1050-3900)	2521 (875-4500)
Mean sperm vials cryopreserved	3 (1-7)	6 (1-12)
Mean sperm concentration (million/mL)	23.6 (0-160)	115.8 (1-338)
Mean sperm motility (%)	49 (24-73)	50 (4-72)

- No complications reported
- Four oocyte cryopreservation patients required additional pain medication post-procedure
- One patients cycle canceled due to poor response

Objective:
To evaluate the safety of ovarian stimulation and assess outcomes of oocyte and sperm cryopreservation in individuals with SCD and TDT prior to gene therapy

Conclusion

- GT for SCD and TDT necessitate gonadotoxic conditioning, which can lead to infertility
- Providing FP is extremely important for patients pursuing curative therapy
- This case series demonstrates that oocyte and sperm cryopreservation is feasible, safe and successful in patients with SCD and TDT
- Special attention should be given to perioperative care and postoperative pain management in patients with SCD