Hematopoietic Stem Cell Gene Therapy for Cystinosis: Updated Results from the Phase 1/2 Clinical Trial

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We will report the results from the phase 1/2 clinical trial (NCT03897361) conducted at the University of California San Diego evaluating the safety and efficacy of CTNS-RD-04, consisting of autologous $CD34^+$ hematopoietic stem cells (HSCs) gene-modified with a lentiviral vector carrying the *CTNS* cDNA in cystinosis. Four patients (ages 20 to 46 years) have been treated with CTNS-RD-04 with follow-up ranging from 1-month to 27 months. CTNS-RD-04 cell doses ranged from 3.63×10^6 to 9.59×10^6 $CD34^+$ cells/kg with vector copy numbers (VCNs; which the number of lentiviral vector per cell) ranging from 0.59 to 2.1 copies/dg. No adverse event related to the drug product and no serious adverse events have been reported. In the 3 first patients with follow-up ranging from 12-months to 26 months, peripheral blood VCN at 12 months post-gene therapy ranged between 0.90 to 2.59. White blood cell cystine was decreased in the 3 treated patients as well as tissue cystine crystals in the skin and rectal mucosa. The 4 patients are no longer taking oral cysteamine. Patient 2 has elected to restart eyedrop cysteamine after a year post-transplant. Updated data will be presented for the four patients infused to date.