# Clinical Trials Data KIT - Document 71

# Autologous CD34+ Hematopoietic Stem Cells Transduced ex Vivo With Elongation Factor 1 Alpha Shortened (EFS) Lentiviral Vector Encoding for the Human ADA Gene

## Clinical Trial: https://clinicaltrials.gov/study/NCT01852071

"eligibilityCriteria": "Inclusion Criteria:\n\n-Children \u2265 1.0 months of age with a diagnosis of ADA-deficient SCID based on A. Decreased ADA enzymatic activity in erythrocytes, leukocytes, skin fibroblasts, or in cultured fetal cells to levels consistent with ADA-deficient SCID as determined by reference laboratory or confirmed ADA gene mutation(s) known to cause disease , AND\n\nB. Evidence of severe combined immunodeficiency based on either:\n\n1. Family history of first order relative with ADA deficiency and clinical and laboratory evidence of severe immunologic deficiency, OR\n2. Evidence of severe immunologic deficiency in subject prior to institution of immune restorative therapy, based on\n\n 1. lymphopenia (absolute lymphocyte count \\<400 cells/mcL) OR absence or low number of T cells (absolute CD3+ count \\<300 cells/mcL) OR\n 2. severely decreased T lymphocyte blastogenic responses to phytohemagglutinin (either \\<10% of lower limit of normal controls for the diagnostic laboratory, \\<10% of the response of the normal control of the day, or stimulation index \\<10)\n\n \* Ineligible for matched sibling allogeneic bone marrow transplantation: absence of a medically eligible HLA-identical sibling, with normal immune function, who may serve as an allogeneic bone marrow donor\n \* Signed written informed consent according to guidelines of the Institutional Review Board (IRB) (UCLA Office of Human Research Protection Program and National Human Genome Research Institute (NHGRI) IRB\n\nExclusion Criteria:\n\n1. Age \u2264 1.0 months Appropriate organ function as outlined below must be observed within 60 days of entering this trial.\n2. Hematologic\n\n 1. Anemia (hemoglobin \\< 10.5 g/dl at \\< 2 years of age, or \\< 11.5 g/dl at \\> 2 years of age).\n 2. Neutropenia (absolute granulocyte count \\<500/mm3.\n 3. Thrombocytopenia (platelet count \\< 150,000/mm3, at any age).\n 4. International Normalised Ratio (INR) or Prothrombin Time (PT) \\> 2 times the upper limits of normal or Partial Thromboplastin Time (PTT) \\> 2.33 times the upper limit of normal (patients with a correctable deficiency controlled on medication will not be excluded).\n 5. Cytogenetic abnormalities on peripheral blood or bone marrow or amniotic fluid (if available).\n 6. Prior allogeneic Hematopoietic Stem Cell Transplant (HSCT) with cytoreductive conditioning\n3. Infectious\n\n a. Evidence of infection with HIV-1, hepatitis B, Hepatitis C, or parvovirus B 19 by DNA Polymerase Chain Reaction (PCR) within 90 days prior to bone marrow harvest. If other infection is present, it must be under control (e.g. stable or decreasing viral load) at the time of screening\n4. Pulmonary\n\n 1. Resting O2 saturation by pulse oximetry \\< 95% on room air.\n 2. Chest x-ray indicating active or progressive pulmonary disease.\n5. Cardiac\n\n 1. Abnormal electrocardiogram (EKG) indicating cardiac pathology.\n 2. Uncorrected congenital cardiac malformation with clinical symptomatology.\n 3. Active cardiac disease, including clinical evidence of congestive heart failure, cyanosis, hypotension.\n 4. Poor cardiac function as evidenced by LV ejection fraction \\< 40% on echocardiogram.\n6. Neurologic\n\n 1. Significant neurologic abnormality by examination.\n 2. Uncontrolled seizure disorder.\n7. Renal\n\n 1. Renal insufficiency: serum creatinine \\>= 1.2 mg/dl, or \\>= 3+ proteinuria.\n 2. Abnormal serum sodium, potassium, calcium, magnesium, phosphate at grade III or IV by Division of AIDS Toxicity Scale.\n8. Hepatic/GI:\n\n 1. Serum transaminases \\> 5 times the upper limit of normal (ULN).\n 2. Serum bilirubin \\> 2 times ULN.\n 3. Serum glucose \\> 1.5 times ULN.\n 4. Intractable severe diarrhea.\n9. Oncologic\n\n 1. Evidence of active malignant disease other than dermatofibrosarcoma protuberans (DFSP)\n 2. Evidence of DFSP expected to require anti-neoplastic therapy within the 5 years following the infusion of genetically corrected cells\n 3. Evidence of DFSP expected to be life limiting within the 5 years following the infusion of genetically corrected cells\n10. Known sensitivity to Busulfan\n11. General\n\n 1. Expected survival \\< 6 months.\n 2. Pregnant.\n 3. Major congenital anomaly.\n 4. Ineligible for autologous HSCT by the criteria at the clinical site.\n 5. Other conditions which in the opinion of the principal investigator and/or co-investigators, contra-indicate the bone marrow harvest, the administration of busulfan, infusion of transduced cells or indicate the patient or patient's parents/primary caregivers inability to follow protocol.",  
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 "sex": "ALL",  
 "minimumAge": "1 Month",  
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"CHILD"  
]

Question:  
Based on the following clinical trial information, would a patient with a KIT gene mutation be eligible for the clinical trial?

Answer:  
No. This trial is specifically for patients with \*\*ADA-deficient SCID\*\*, confirmed by either low ADA enzyme activity or ADA gene mutations. While the patient in question \*does\* have a gene mutation, it's a KIT gene mutation, \*\*not\*\* an ADA gene mutation. Therefore, they would not be eligible for this particular trial.