FACTOR IX VARIANTS WITH ENHANCED BIOLOGICAL FUNCTIONS

Published date: Feb. 27, 2018

Technology description

Market Need

Hemophilia B, also known as Christmas disease, is an x-linked genetic disorder caused by missing or defective Factor IX (FIX), a clotting protein. It is estimated that hemophilia occurs 1 in every 5000 births with 20,000 estimated to have hemophilia in the US alone. The severity of decrease in FIX levels in Hemophilia B patients varies from mild to severe and in severe cases, which represent approximately 60% of cases, the decreased levels of FIX (<1% of FIX in blood) causes patients to experience frequent spontaneous bleeding episodes and bleeding episodes after injuries. Currently, treatment is mainly the delivery of recombinant FIX product, which is delivered intravenously through a vein the arm or port in the chest. In some severe cases, patients with severe hemophilia may undergo prophylactic FIX delivery to prevent bleeding. These recombinant FIX treatments are costly and require multiple visits to a specialist. Furthermore, because of the short half-lives of the recombinant factors have patients typically undergo multiple injections at high doses. Thus, there is a significant need for treatments that would decrease these burdens on patients and on the healthcare system.

Technology Overview

One focus of the Arruda and Camire labs is the development of AAV vectors that encode clotting factors, including FIX. These investigators have jointly developed nine improved variants of FIX, deliverable via gene therapy approaches, which show up to 17 fold increase in specific activity in vivo when compared to wildtype FIX. These variants were discovered through a screening strategy to identify regions of the protein that were spare from loss of function mutations and regions that could potentially bind to activated factor VIII, an important part of thrombin and clot formation.

Advantages

- Higher specific activity compared to wildtype FIX
- Gene therapy approach will result in less frequent visits to hospital for treatment

Institution

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