OT-58 as an Enzyme Replacement Therapy for Patients With Cystathionine Beta-Synthase Deficient Homocystinuria (CBSDH)

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Cystathionine Beta-Synthase (CBS) Deficient Homocystinuria (CBSDH) is a rare autosomal-recessive metabolic condition characterized by an excess of homocysteine (Hcy) in the plasma, tissues and urine. It is due to reduced or absent activity of the Cystathionine Beta-Synthase (CBS) enzyme, and is also known as classical homocystinuria. The symptoms associated with CBSDH are variable in severity and time of onset across patients. Some affected individuals may have mild signs of the disorder; others may have multi-systemic involvement including potentially life-threatening complications. CBSDH can affect many different organ systems of the body; the four most commonly involved are the eyes, central nervous system, skeleton, and the vascular system.

The current approaches to treatment of CBSDH patients include a highly restrictive diet and use of dietary supplements. Lifetime compliance with this diet is poor. OT-58 represents a novel therapeutic approach that incorporates the use of a modified version of the native, human CBS (hCBS) enzyme. The goal of treatment is to introduce the CBS enzyme into circulation, resulting in reduced Hcy levels, increased cystathionine levels and normalized cysteine (Cys) levels.