Glycerol phenylbutyrate granted orphan drug designation for treatment of STXBP1 developmental and epileptic encephalopathy

Stockholm, October 20, 2023 – Immedica today announces that the European Commission has granted an orphan designation for glycerol phenylbutyrate in STXBP1 developmental and epileptic encephalopathy (STXBP1-DEE), following positive opinion from EMA's Committee for Orphan Medicinal Products (COMP).

Orphan designation is granted to medicines that treat, prevent, or diagnose a life-threatening or chronically debilitating rare disease, with a prevalence in the EU of below 5 in 10,000, and with either no satisfactory method of diagnosis, prevention or treatment or with significant benefit to those affected by the disease.

About STXBP1-DEE
STXBP1-DEE is a rare genetic neurological disorder, affecting children and adults, due to a defect in the STXBP1 gene. The condition is chronically debilitating due to neurodevelopmental abnormalities, epilepsy, and motor and behavioural disturbances. There are no curative, disease-altering, or specific therapies available for STXBP1-DEE patients. Current treatments are therefore symptomatic and supportive. As no medicines are approved specifically for STXBP1-DEE, seizure control via treatment with anti-epileptic drugs represents the most common treatment offered to patients. Disease management for developmental delay, cognitive dysfunction and intellectual disability consists of physiotherapy, occupational therapy, and speech language therapy. The estimated incidence is 1:30 000.

About glycerol phenylbutyrate
Glycerol phenylbutyrate, is the active ingredient in a medicine currently being used for treatment of urea cycle disorders including deficiencies of carbamoyl phosphate synthetase I (CPS), ornithine carbamoyltransferase (OTC), argininosuccinate synthetase (ASS), argininosuccinate lyase (ASL), arginase I (ARG) and ornithine translocase deficiency hyperornithinaemia-hyperammonaemia homocitrullinuria syndrome (HHH) that cannot be managed by dietary protein restriction and/or amino acid supplementation alone. There is some preliminary non-clinical and clinical data suggesting glycerol phenylbutyrate may benefit patients with STXBP1-DEE. Glycerol phenylbutyrate is not approved for treatment of STXBP1-DEE.

About Immedica
Immedica is pharmaceutical company, headquartered in Stockholm, Sweden, focused on the commercialization of medicines for rare diseases and specialty care products. Immedica's capabilities cover marketing and sales, compliance, pharmacovigilance, quality assurance, regulatory and medical affairs as well as market access. Serving patients in more than 50 countries, Immedica is fully dedicated to helping those living with diseases which have a large unmet medical need.

Immedica's therapeutic areas are within genetic & metabolic diseases, hematology & oncology and specialty care.

Immedica was founded in 2018 by the investment company Impilo and Buy-in-Management. Today Immedica employs more than 100 people across Europe and the Middle East.

For more information visit www.immedica.com

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