Case report: First time use of Lomitapide in United Arab Emirates in Autosomal Recessive Familial Hypercholesterolemia – A RARE DISEASE

Background

Autosomal recessive hypercholesterolemia (ARH) is a very rare monogenic disorder affecting less than 1 in 1000,000 people and is characterized by very high levels of low-density lipoprotein cholesterol (LDL-C), leading to aggressive and premature atherosclerotic cardiovascular disease if left untreated. Lomitapide, an inhibitor of microsomal triglycerides transferase protein (MTP), has been proven to decrease LDL-C levels by almost 50% as an adjunct to other lipid-lowering medications in homozygous familial hypercholesterolemia.

Clinical report

We report a 42 year Emirati female referred to the Lipid Clinic Al Ain in 2017; genetic test confirmed diagnosis of Autosomal Recessive Hypercholesterolemia *LDLRAP1*: Homozygous c.604delTinsCC p.(Ser202fs) pathogenic sequence variant. Initial LDL-C of 12 mmol/L came down to 7- 7.9 mmol/L with Rosuvastatin 40 mg and Ezetimbe 10 mg daily. Addition of PCSK9 inhibitor Evolocumab decreased LDL-C to 4.0 mmol/L. With dietary counselling to reduce fat intake to less than 15-20 % in daily calories, LDL-C came down to 3.3-3.6mmol/L. Upon availability of Lomitapide in the United Arab Emirates this medicine was added to this patient's treatment regimen at an initial dose of 5 mg for 4 weeks followed by 10 mg daily and the LDL-C came down to 1.5 mmol/L with almost 50 % further reduction. No adverse effects were noted.

Conclusion

Lomitapide is an oral microsomal triglycerides transferase protein (MTP) inhibitor approved for the treatment of Homozygous Familial Hypercholesterolemia and appears to be a realistic option for this patient as an effective and well-tolerated adjunct therapy. Further studies are required to establish the role of this medicine in the treatment of this rare disease.

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