EA067

MTOR-INHIBITOR SIROLIMUS IN THE TREATMENT OF CONGENITAL LYMPHATIC MALFORMATIONS.

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Introduction

Congenital Isolated Lymphatic Malformations (ILM) are disorders of lymphangiogenesis. Recent studies have correlated mTOR-pathway gene somatic mutations, in PIK3CA gene, in the aetiology of the disorder, including them in the spectrum of PIK3CA related Overgrowth Spectrum disorders (PROS).

Purpose

Treatment of ILM with mTOR inhibitor sirolimus and monitor of the clinical response according to a protocol submitted at Attikon University Hospital.

Material & Method

Parents were informed, inclusion and exclusion criteria were studied and after obtained consent, extensive work-up was performed and patients were enrolled and commenced on sirolimus, 0.8mg/m2 BID. Monitoring of SRL levels at low threshold (2-6ng/l), frequent blood tests and clinical assessments were performed, along with a 6-monthly MRI to study the effect of the treatment. Five paediatric patients with ILM were enrolled presenting the following lesions: gigantic right thoracocervical lesion, left extra-abdominal lesion, right posterior thoracic lesion, diffuse intrabdominal lesion and a right orbital and eyelid ILM.

Results

In the case of the gigantic ILM, SRL was commenced at 3months of age and a significant reduction of ILM size was noted. A sclerotherapy was also performed in combination and she is currently off treatment with no regression. In the following 3 cases SRL led to a significant reduction of lesion size. Orbital ILM was treated in combination with sclerotherapy with moderate results. No significant adverse events were noted.

Conclusions

SRL is an aetiologic treatment that should be considered in patients with ILM.