



HAEMANGIOMAS AND VASCULAR MALFORMATIONS

SIROLIMUS IN BLUE RUBBER BLEB NAEVUS SYNDROME: A SYSTEMATIC REVIEW

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Introduction: Blue rubber bleb naevus syndrome is an uncommon disorder characterised by multifocal venous malformations involving the skin, gastrointestinal tract and less frequently viscera. Currently, there exists no curative therapy nor guidelines regarding best management of this multisystem disease. Sirolimus has recently been employed in a range of paediatric vascular anomalies, including blue rubber bleb naevus in small volume case reports

Objective: To systematically review and evaluate the evidence regarding sirolimus treatment for blue rubber bleb naevus syndrome.

Materials and Methods: A literature search of Medline, Embase, CINAHL, SCOPUS and Google Scholar was conducted. Publications reporting the use of sirolimus for any period of time in patients with a clinical diagnosis of blue rubber bleb naevus were included. Articles that reported patients with vascular malformations without specification of subtype or with insufficient clinical detail were excluded.

Results: Of 46 articles identified through the search strategy, 17 studies reporting 23 patients met inclusion criteria. Sirolimus regimens varied greatly; a reflection of the broad range in age and heterogeneity of treating institutions. Some treating physicians based dosage on body mass and some on body surface area. Titration of dose to trough levels and Pneumocystis prophylaxis was also variably employed.

22/23 patients observed an improvement in either cutaneous or gastrointestinal disease. 17/18 patients noted an improvement in gastrointestinal disease where this was reported. 21/22 patients noted an improvement in cutaneous disease where this was reported. Sirolimus was well tolerated in all patients but one patient who required treatment cessation.

Conclusions: The results of this systematic review demonstrate that there is strong cumulative evidence for the role of sirolimus in managing patients with blue rubber bleb naevus syndrome. Depending on the degree of morbidity, it may be considered first-line therapy.

