RESEARCH SIMPLIFIED

Title: Clinical Profile of Overgrowth Syndromes Consistent with PROS (*PIK3CA*-Related Overgrowth Syndromes)—A Case Series

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Research Simplified By: Lydia Mathew

Short biography

I work as Assistant Professor in the department of dermatology at Christian Medical College, Vellore, India. I completed my under-graduate medical studies in 2006 and my post graduate training in dermatology in 2013 at the same institute. As a consultant I am involved in the out-patient and in-patient care of patients with dermatological diseases and train under-graduate and post-graduate students in the specialty. My interests include paediatric dermatology specifically cutaneous vascular anomalies and overgrowth syndromes. I got intrigued with PROS after encountering a patient with MCAP syndrome the diagnosis of which was made by the dermatology team. Since then, we have been working as a team with the interventional radiologists, the paediatric and plastic surgeons for the management of such patients. I am currently involved in a research on PIK3 mutation in lesional tissues of patients with suspected PROS. Once we establish tests for mutation analysis in PROS, we hope to offer diagnostic services which is currently lacking in India and thereby direct appropriate management for such patients.

Research Simplified Summary:

Background - PROS is a condition with unusual and progressive growth of certain body structures and sometimes of the brain due to a spontaneous change in the genetic structure of tissues. It may be obvious when the child is born and becomes evident as the child grows. Birth marks on the skin along with other clinical features help doctors arrive at a probable diagnosis. Based on their unique combination of features they might be referred to by specific names such as fibroadipose overgrowth (FAO), congenital lipomatous overgrowth, vascular malformations, epidermal nevi, spinal, and skeletal anomalies syndrome (CLOVES) to name a few.

Aim of the study – To describe the clinical features of patients suspected to have PROS from the Indian subcontinent

Study group – We looked at medical records of 15 patients with features of excessive growth of body parts who presented to us over 70 months from July 2012 to April 2018 at the department of dermatology at a referral hospital in South India.

Results – There were nine males and six females in our study group ranging from ages 8 months to 73 years. Among them, five patients had features of FAO, five had CLOVES, two had megalencephaly capillary malformation (MCAP) syndrome, one each had features of hemi-hyperplasia multiple lipomatosis (HHML) and congenital infiltrative diffuse lipomatosis (CDIL). One patient did not have specific features of any of the defined entities. The overgrowth was slowly progressive in all patients and affected the limbs in thirteen patients, one half of the face in two patients and the head in three patients. Eleven patients had altered formation of their toes or fingers. Fatty growths on the body were seen in seven patients while one patient had reduced fat on one half of the face.

We found anomalies in blood vessels of the skin in 60 % (nine patients) and epidermal nevi (benign growth of cells in the outermost layer of the skin) in 46.6 % (seven patients) the frequencies of which were higher than that reported previously in published medical literature. Port wine stains were the commonest vascular birthmarks. Other vessels such as the veins and the lymphatics were also affected in a few. The nevi on skin and the vascular birthmarks were sometimes noted on the overgrown body part and extending beyond. Other features on the skin included increased hair growth of overgrown areas, brownish colored birthmarks (café au lait macules) in a few and unusual features such as groups of skin tags near the overgrown upper limb in one patient and near the overgrown buttock in another patient,

streaky pale patches along the neck and small linear soft tissue growths in the mouth. None of the skin lesions posed any danger to the patient.

Other systems were also affected such as the brain (26.6%, four patients) who had either developmental problems or seizures. Few of our male patients (33.3%, three of nine male patients) had surgically correctible developmental problems of the genital organs. Skeletal issues including of the spine was seen in one patient with CLOVES. None of our patients had any internal cancerous growths till the study period and were otherwise healthy.

Conclusion – By performing this study we were able to provide more data on the varied clinical spectrum of patients with PROS from the Indian sub-continent. Testing for mutations in affected tissues is the recommended diagnostic test which may not always be accessible to patients from countries such as ours. In such situations, careful clinical examination by doctors from multiple specialties including that by a dermatologist will help suspect this condition. Attention to the development of genito-urinary organs and the brain as is screening for malignancies is recommended.