Yan-Hua Liang, J Clin Exp Dermatol Res 2014, 5:2
http://dx.doi.org/10.4172/2155-9554.S1.015

OMICS Group Conferences
Accelerating Scientific Discovery

4th International Conference on
Clinical & Experimental Dermatology
April 14-16, 2014 Hilton San Antonio Airport, TX, USA

Newly identified phenotypes in a FIP1L1/PDGFRA-associated pediatric HES patient: Thrombocytosis, mHPA, young stroke and blindness

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Background: Hypereosinophilic syndromes (HES) constitute a heterogeneous group of disorders, defined as persistent and marked blood eosinophilia (>1.5×109/L, ≥six consecutive months) associated with eosinophil-induced organ damages, where other causes of hypereosinophilia have been excluded. The clinical manifestations of HES are variable from one patient to another, depending on target-organ infiltration by eosinophils. Although virtually any tissue at any age can be concerned, complications arise most frequently in the skin, heart, lungs, and nervous system at the age of fifties. Pediatric HES is very rare, with only 33 case reports in literature.

Methods: A 9-year old Chinese girl of multi-organ defects was included in this study. Chest X-ray, computer tomography, electrocaardiogram, electroencephalogram, and doppler ultrasound were used for full examination. Laboratory tests were obtained from various items, including skin immunohistology, bone marrow aspiration, FACS analysis, trace element analysis, inborn errors of metabolism, karyotype analysis and nest-PCR for FIP1L1/PDGFRA.

Results: Informative data revealed a diagnosis of HES with dysfunctions of multiple organs. Nest-PCR identified the fusion fragment between FIP1L1 and PDGFRA gene. These HES patient present novel phenotypes, such as thrombocytosis, mild hyperphenylalaninemia (mHPA), weak eyesight and blindness, and young ischaemic stroke. Disease conditions were significantly improved by imatinib treatment.

Conclusion: This HES case was characterized by the earliest onset with novel phenotypes including blindness, thrombocytosis, mHPA, and young stroke. Imatinib was effective to resolute these problems.

Biography

Yanhua Liang, M.D., Ph.D., Associate Professor in the Department of Dermatology, Nanfang Hospital at Southern Medical University, China, received his Ph.D. in Dermatology in 2007. He had his 2-year postdoctoral training at The Jackson Laboratory USA before jointing the faculty of Yale University School of Medicine in 2010. He has identified *CYLD1* gene as the disease gene of multiple familiar trichoepithelioma, and *XBP1* as genetic risk factor for vitiligo in Chinese Hans. He has done in-depth research to understand the biological function of SHARPIN. Except for molecular basical studies, he has also developed new biological materials and techniques for transdermal gene delivery, and modified *in vivo* models for hair follicle reconstitution. He has established a translational skin research program with major focus on skin tumor and genodermatitis. Dr. Liang is the author of more than 50 peer reviewed publications with more than 600 citations, joined the editorial board of *J Dermatol Clin* Res, and serves as the reviewer of nine renowned journal such as *J Invest Dermatol.*

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