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Abstract

Genetic Discrepancy in Chediak-Higashi syndrome Patients

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Abstract:

Adenosine deaminase (ADA) deficiency is one of the primary immunodeficiency diseases, that affects the immune system through accumulated metabolites of purine metabolism. It is an autosomal-recessive disorder with an estimated incidence around 0.5 per 100,000 live births. Patients with severe form usually present in infancy where the accumulation of the toxic metabolites; deoxyadenosine and deoxyadenosine triphosphate (dAXP), causing defective differentiation and function of immune cells; T, B, and natural killer (NK) leading to severe combined immunodeficiny. Those patients have significant recurrent invasive infections that usually the cause of thier deaths early in life, but if they servive, they usually have a lot of long term complications. We previously reported in multicenter studay, Hematopoietic stem cell transplantation (HSCT) is the ideal treatment of choice if an human leukocyte antigen (HLA)- match sibling donor is avialable, however, alternatively HLA-matched unrelated donor (MUD) transplant could be used. The enzyme replacement therapy approach using polyethylene-glycol–modified bovine ADA (PEG-ADA), if available, could save life of those patients, but it is very expensive and it was observed that the immune system deteriorates over time. Gene therapy to treat ADA defecient patients, is currentlly an opition if HLA matched donor is not avialable for HSCT. Gene therapy, using autologous CD34+-enriched cell fraction that contained CD34+ cells transduced with a retroviral vector encoding the human ADA complementary DNA sequence (GSK2696273) had been showen to be effective and safe.

Biography:

Dr. D. ALZahrani has long experience in diagnosing and managing primary immunodeficiency diseases. Dr. D. ALZahrani received bachelor degree of medicine and surgery form King Saud University in 1994. He completed pediatric residency training program at the University

of B.C. Vancouver, Canada. He received American Board of Pediatrics in October 2003. Then, he had Allergy and Clinical Immunology Fellowship for 2-years at McMaster University. He got Fellowship training in Bone Marrow Transplant and Primary Immunodeficiency at Hospital for Sick Children, University of Toronto, Canada. He received American Board of Allergy and Immunology (ABAI) in Oct 2006. Since 2007 till present, works as Consultant allergy, immunology and BMT at King Abdulaziz medical City – WR, and King Saud Bin Abdulaziz University for health sciences, Jeddah Saudi Arabia.

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