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#### **REVIEW ARTICLE**

### Molecular nature of alpha-globin genes in the Saudi population

Characteristics	PIB, n=32	Controls, n=33	P-value
Age, years	10.2 (1.5 - 16)	10 (1.5 - 16)	0.18
Gender, male	56%	58%	
Follow up duration, months	33 (6 - 78)	42 (12 - 72)	
Deceased donors	56%	52%	
Time period	January 2003 to December 2011	April 2005 and May 2011	

Molecular studies on the alpha-globin genes in Saudi population reveals that the HBA2 gene is replaced by a unique HBA12 gene convert in 5.7% of the population. This review by Borgio summarizes the existence of 32 different varieties of  $\alpha$ -globin genotypes among the population. Borgio insists the necessity of sequence based testing systems to identify the sequence defects in HBA2, HBA1, HBA12 and ATRX genes to enhance the existing premarital screening program in the region, and thus may reduce the number of newborns with thalassemia.

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#### **ORIGINAL ARTICLES**

#### Can pre-implantation biopsies predict renal allograft function in pediatric renal transplant recipients?

Pre-implantation kidney transplant biopsies in children in this study provide important baseline information regarding the transplant. In this study by Kari et al, normal kidney transplant biopsies are obtained in 41% of children who underwent kidney transplantation. The results from all of the biopsies provide invaluable information, which can assist doctors in understanding the implications of subsequent events and medical treatment. Compared with a control group, children with histological changes in pre-implantation biopsy, such as pre-existing chronic vascular changes are more likely to have worse prognosis, with delayed graft function and/or renal allograft loss.

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## Favorable therapeutic response of osteoporosis patients to treatment with intravenous zoledronate compared with oral alendronate

Al-Bogami et al report a favorable response in osteoporosis patients to intravenous compared with oral bisphosphonates. In this prospective study, the authors evaluate the efficacy of zoledronate and alendronate on improving bone mineral density (BMD) in 234 patients. Using DEXA, the authors reveal that while lumbar spine BMD after 2 years increased by 5.69% in patients treated with zoledronate and 3.58% in patients treated with alendronate, BMD in the hip decreased with alendronate but increased with zoledronate by 0.78%. The study concludes that zoledronate provides advantages in compliance, reduced side effects, and higher efficacy in halting osteoporosis than alendronate.

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#### **CASE REPORT**

#### Glutaric aciduria type 1 as a cause of dystonic cerebral palsy



Follow up magnetic resonance imaging brain study after 3 months. Axial T2 weighted image showing reduced size (atrophy) of basal ganglia and progressive bilateral fronto-temporal atrophy.

Glutaric aciduria type 1 (GA1) is an inherited inborn error of metabolism caused by a deficiency of the enzyme glutaryl Co-A dehydrogenase (GCDH). This report by Mohamed et al discusses a 14-month-old Saudi boy with GA1 who presented with severe dystonia and was misdiagnosed as cerebral palsy (CP). His DNA analysis confirmed homozygosity for a mutation in the GCDH-coding gene (c.482G>A;p.R161Q). This case alerts pediatricians to consider GA1 as a differential diagnosis of children presenting with dystonic CP, as early intervention prevents neurologic disabilities. Also, all children with neurologic symptoms of unknown origin, such as CP should undergo work-up for inborn errors of metabolism.

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