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Research Article

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Cystinosis - Pathophysiology

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Cystinosis is a rare autosomal recessive lysosomal storage disorder affecting 1 in 100,000 – 200,000 live births. It is caused by a mutation in the Cystinosin (CTNS) gene, a cystine-proton cotransporter, the absence of which results in intra-lysosomal accumulation of cystine. Kidneys are affected first, presenting as Fanconi syndrome in infancy, followed by widespread ...

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Analysis of induction and maintenance immunosuppression choices in the US during the first year post kidney transplant for patients over 70

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Author(s): Amy H White*, John Hunton, Saleema Karim, Allison Wells, Hanna Jensen, Darby Derringer, Misha Karr, Sathyanand Kumaran and Lyle Burdine

Rates of kidney transplantation in patients over 70 years of age have steadily increased over the last 20 years, however age-appropriate immunosuppression regimens in the elderly remain unclear. Investigators utilized the SRTR database to evaluate elderly kidney transplant recipients' outcomes against a younger population. Post-transplant outcomes measured at an app ...

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Case Report

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Ochoa syndrome: An overlooked diagnosis – A case report

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Author(s): Mário Nicolau Barros Jacobino*, Patrícia Barros Aquino Jacobino, Matheus Saraiva Lopes and Francisco Arisneto Avelino Fontenele Júnior

The Urofacial Syndrome or Ochoa is a very rare clinical condition, and is unknown by a large part of the medical community; it is characterized by an inverted facial expression, resulting from abnormal contraction of facial and ocular muscles, especially when smiling, in addition to the presence of urinary abnormalities. Patients with this syndrome are at a higher ris ...

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