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Our **case** has received a successful **renal** transplant from brother which is working well for the last 3 years without any evidence of recurrence. Another interesting feature of our **case** was the strong family history of unrelated other **renal** genetic disorder autosomal dominant adult **polycystic kidney disease** ...

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autosomal dominant **polycystic kidney disease** (ADPKD) is a heterogenetic disorder caused by **mutations** in(2, 5) PKD1 (chromosome region 16p13.3) encodes for polycystin-1 reported in 85% of cases may cause more severe **disease**, with mean age of end-stage **renal disease** (ESRD) onset 58 years PKD2 (chromosome region 4q21) encodes for polycystin-2

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Conclusions . We present a male patient with **OFD1 mutation** who lacks the classic **OFD1 phenotype** who presented with **end-stage renal disease** without evidence of **polycystic changes** within the **kidneys**.

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renal tubular cysts develop in several inherited human disorders. Among these, the **polycystic kidney** diseases (PKD) are one of the leading causes of end-stage **renal disease** in children and adults ().Autosomal dominant **polycystic kidney disease** (ADPKD) occurs in 1:1,000 individuals, primarily as the result of **mutations** in one of **two** genes, PKD1 or PKD2 (81, 150–152).

Cited by: 237 **Author:** Lisa M. Guay-Woodford
Publish Year: 2003

Name of Journal: *World Journal of Clinical Cases*

Manuscript NO: 52524

Manuscript Type: CASE REPORT

OFD1 mutation induced renal failure and polycystic kidney disease in a childhood male twins in China

Hong-Wen Zhang, Bai-Ge Su, Yong Yao

Abstract

BACKGROUND

Oral-facial-digital syndrome type 1 (OFD1) is a rare ciliopathy mainly with an X-linked dominant inheritance, which is caused by mutations in the *OFD1* gene. The encoding protein of OFD1 is located within centrosomes and basal bodies

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Management of delivery of a fetus with autosomal recessive ...

https://jmedicalcasereports.biomedcentral.com/articles/10.1186/s13256-019-2293-3

Background. Autosomal recessive **renal polycystic kidney disease** occurs in 1 in 20,000 live births. It is caused by **mutations** in both alleles of the PKHD1 gene. Management of deliv

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