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Two Hereditary Renal Diseases In The Same Algerian Family: A Rare Case Report

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Abstract:

Introduction: Primary hyperoxaluria type 1 (PH1) is the most frequent and severe form of primary hyperoxalurias. It is a rare autosomal recessive disease caused by an enzymatic deficiency in alanine-glyoxylate aminotransferase (AGT). Autosomal dominant polycystic kidney disease (ADPKD) is a hereditary condition characterized by the development in adulthood, but in 72% of cases during the second decade, of multiple cysts in the kidneys and very often in the liver. It is the most common hereditary renal disease worldwide.

Patients and Methods: Patient M.S., an 11-year-old boy from a first-degree consanguineous marriage, only son of two siblings originating from northern Algeria, with a family history of a sister and mother affected by ADPKD, diagnosed at 12 and 20 years respectively. The child consulted for bilateral renal colic associated with recurrent urinary tract infections beginning at the age of two years with stone expulsion. Clinical examination revealed a child in good general condition, no growth retardation, normal blood pressure at 110/70 mmHg, polyuria greater than 3 L/day. Urinalysis showed morning pH at 5.5, the rest without abnormalities. Laboratory blood and urine tests including complete blood count, renal function, serum electrolytes, calcium-phosphorus, liver and lipid profiles were normal. Calciuria was 1.08 mmol/L and hyperoxaluria was 449 mmol/L. The crystalline composition of expelled stones was whewellite (calcium oxalate monohydrate). Morphological assessment with plain abdominal X-ray and abdominopelvic ultrasound revealed nephrocalcinosis. Molecular analysis was performed by direct sequencing of exons 1, 4, 7, and 10 of the AGXT gene. The proband was homozygous for the c.853C>T (p.Ile244Thr) mutation, known as the "Maghrebian" mutation [Nagara et al., 2013; Belhaj et al., 2011]. Both parents were heterozygous for this mutation, and the sister did not carry the mutation. The child was started on diuretic therapy and pyridoxine at 500 mg/day, with significant improvement in oxalemia and oxaluria levels measured one year later.

Discussion and Conclusion: This is a first-degree consanguineous family presenting two hereditary renal diseases that can lead to extrarenal replacement therapy. A detailed literature review on the Maghrebian mutation of PH1 and the studied populations [Nagara et al., 2013; Belhaj et al., 2011], as well as the rare observations of an association of two genetic renal diseases described in the same family, was performed. The dual hereditary renal involvement in this family posed the problem of genetic counseling and living-related renal transplantation. As the mother and sister have ADPKD, they cannot serve as kidney donors. The father remains the only potential donor. The development of cadaveric organ transplantation in our country remains the only hope for this family.

Key words: primary hyperoxaluria type 1; autosomal dominant polycystic kidney disease; maghrebian mutation; agxt gene; nephrocalcinosis; genetic counseling; renal transplantation

Introduction:

Primary hyperoxaluria type 1 is the most frequent and severe form of primary hyperoxalurias [1, 2]. It is a rare autosomal recessive disease caused by a deficiency of the hepatic peroxisomal enzyme alanine-glyoxylate aminotransferase, encoded by the AGXT gene located on chromosome 2q37.3 [1, 2]. This enzyme deficiency leads to overproduction of oxalate, resulting in hyperoxaluria, recurrent urolithiasis, nephrocalcinosis, and progressive renal failure [1, 2]. Primary hyperoxaluria type 1 is responsible for 1% to 2% of pediatric kidney failure cases [3, 4]. The prevalence of primary hyperoxaluria type 1 is estimated at 1 to 3 per million population, with a higher frequency in certain populations, particularly in North Africa where the "Maghrebian" mutation (c.853C>T, p.Ile244Thr) is prevalent [1, 2].

Autosomal dominant polycystic kidney disease is a hereditary condition characterized by the development in adulthood, but in 72% of cases during the second decade, of multiple cysts in the kidneys and very often in the liver [Chebib et al., 2025]. It is the most common hereditary renal disease worldwide, with a prevalence of 1 in 400 to 1 in 1000 live births [5, 6]. Autosomal dominant polycystic kidney disease accounts for 5% to 10% of kidney failure in the US and Europe and is primarily caused by pathogenic variants in the PKD1 (78%) or PKD2 (15%) genes [5]. Approximately 50% of individuals with ADPKD require kidney replacement therapy by 62 years of age [5].

The coexistence of two hereditary renal diseases in the same family is exceptional. We report the case of an Algerian family presenting both primary hyperoxaluria type 1 in a child and autosomal dominant polycystic kidney disease in two other family members. This rare association raises complex issues of genetic counseling and renal transplantation.

Case Report:

Patient History:

Patient M.S., an 11-year-old boy from a first-degree consanguineous marriage, was the only son of two siblings originating from northern Algeria. The family history revealed that his sister and mother were affected by autosomal dominant polycystic kidney disease, diagnosed at 12 and 20 years respectively. The father was healthy.

Clinical Presentation:

The child consulted for bilateral renal colic associated with recurrent urinary tract infections beginning at the age of two years, with expulsion of stones. Clinical examination revealed a child in good general condition, with no growth retardation. Blood pressure was normal at 110/70 mmHg. Polyuria greater than 3 L/day was noted. Urinalysis showed morning pH at 5.5, with no other abnormalities.

Laboratory Findings:

Laboratory blood and urine tests revealed normal complete blood count, renal function, serum electrolytes, calcium-phosphorus levels, liver and lipid profiles. Calciuria was 1.08 mmol/L. Hyperoxaluria was 449 mmol/L. The crystalline composition of expelled stones was whewellite (calcium oxalate monohydrate).

Imaging Findings:

Morphological assessment with plain abdominal X-ray and abdominopelvic ultrasound revealed nephrocalcinosis.

Molecular Analysis:

Molecular analysis was performed by direct sequencing of exons 1, 4, 7, and 10 of the AGXT gene. The proband was homozygous for the c.853C>T (p.Ile244Thr) mutation, known as the "Maghrebian" mutation [1, 2]. Both parents were heterozygous for this mutation, and the sister did not carry the mutation. This finding confirmed the diagnosis of primary hyperoxaluria type 1.

Treatment:

The child was started on diuretic therapy and pyridoxine at 500 mg/day [Adam et al., 1993-2026]. Significant improvement in oxalemia and oxaluria levels was measured one year after treatment initiation.

Discussion:

Primary Hyperoxaluria Type 1:

Primary hyperoxaluria type 1 is a rare autosomal recessive disorder caused by deficiency of the hepatic peroxisomal enzyme alanine-glyoxylate aminotransferase [1, 2]. This enzyme deficiency leads to overproduction of oxalate, which is excreted in the urine and forms calcium oxalate crystals, resulting in recurrent urolithiasis, nephrocalcinosis, and progressive renal failure [1, 2]. The disease typically presents in childhood, with a median age of onset of 5 years [1, 2].

The "Maghrebian" mutation (c.853C>T, p.Ile244Thr) is a founder mutation prevalent in North African populations, particularly in Algeria, Morocco, and Tunisia [1, 2]. This mutation accounts for a significant proportion of PH1 cases in the Maghreb region [1, 2]. Studies have identified the p.I244T mutation as the most predominant mutation in Tunisian patients, along with the p.G190R mutation [1, 2]. Homozygosity for this mutation is associated with a severe phenotype and early onset of end-stage renal disease [1, 2]. In our patient, the early onset of symptoms at the age of two years and the presence of nephrocalcinosis suggest a severe phenotype.

Pyridoxine (vitamin B6) is the mainstay of medical therapy for primary hyperoxaluria type 1 [7]. Pyridoxine acts as a cofactor for alanine-glyoxylate aminotransferase and can increase residual enzyme activity in some patients [8]. Approximately 30% to 50% of patients with PH1 respond to pyridoxine therapy [7]. In our patient, pyridoxine at 500 mg/day led to significant improvement in oxalemia and oxaluria levels, confirming a favorable response.

Autosomal Dominant Polycystic Kidney Disease:

Autosomal dominant polycystic kidney disease is the most common hereditary renal disease worldwide, with a prevalence of 1 in 400 to 1 in 1000 live births [5, 6]. It is characterized by the development of multiple cysts in the kidneys and other organs, particularly the live [5]. The disease typically presents in adulthood, with most persons diagnosed between the ages of 27 and 42 years [5]. Over 90% of patients older than 35 years have hepatic cysts [5]. Hypertension affects 70% to 80% of patients with ADPKD, and approximately 9% to 14% develop intracranial aneurysms [5].

In our family, the mother and sister were diagnosed with ADPKD at 20 and 12 years respectively. The presence of this disease in two generations is consistent with autosomal dominant inheritance.

The Maghrebian Mutation:

The "Maghrebian" mutation (c.853C>T, p.Ile244Thr) is a founder mutation in the AGXT gene prevalent in North African populations [1, 2]. This mutation results in an amino acid substitution that affects the enzyme's function and stability [1]. Homozygosity for this mutation leads to the complete absence of alanine-glyoxylate aminotransferase activity and severe disease [1].

In our patient, homozygosity for this mutation was confirmed by molecular analysis. Both parents were heterozygous carriers, consistent with the autosomal recessive inheritance pattern. The sister, who did not carry the mutation, is not at risk of developing primary hyperoxaluria type 1.

Association of Two Hereditary Renal Diseases in the Same Family:

The coexistence of two hereditary renal diseases in the same family is extremely rare. A review of the literature identified only a few cases of families with both primary hyperoxaluria type 1 and autosomal dominant polycystic kidney disease [9]. The presence of two independent hereditary diseases in the same family raises complex issues of genetic counseling and transplantation.

In our family, the mother and sister with ADPKD cannot serve as kidney donors for the child with PH1, as they are at risk of progressive renal disease themselves. The father, who is healthy and a heterozygous carrier for the PH1 mutation, remains the only potential living donor. However, heterozygous carriers for PH1 may have a higher risk of developing hyperoxaluria and nephrolithiasis, and careful evaluation is necessary [10].

Implications for Genetic Counseling

The presence of two hereditary renal diseases in the same family highlights the importance of genetic counseling [7, 10]. Genetic counseling should include:

- Risk assessment: The risk of transmitting PH1 and ADPKD to offspring should be explained to the family [10].
- Carrier testing: Family members at risk of being carriers should be offered testing [10].
- Prenatal diagnosis: For families with known mutations, prenatal diagnosis should be offered [8].
- Preimplantation genetic diagnosis: For families at high risk, preimplantation genetic diagnosis may be considered [8].

Implications for Renal Transplantation

Renal transplantation is the treatment of choice for patients with PH1 who reach end-stage renal disease [1, 3, 4]. However, transplantation in patients with PH1 carries specific challenges [3, 4].

- Risk of recurrence: Oxalate deposits in the transplanted kidney can lead to recurrence of nephrocalcinosis and graft loss [3, 4].
- Need for combined liver-kidney transplantation: Liver transplantation restores hepatic AGT enzyme activity and is the only curative treatment for PH1 [3, 4]. The native liver must be removed to avoid ongoing pathologic oxalate production [4].
- Living-related donation: As discussed above, living-related donation from heterozygous carriers is controversial [10].

In our family, the development of cadaveric organ transplantation in Algeria remains the only hope for the child. The father, as the only potential living donor, would require careful evaluation to assess the risk of transmission of the disease [3, 4, 10].

Conclusion:

We report the rare case of an Algerian family presenting two hereditary renal diseases in the same family: primary hyperoxaluria type 1 in a child and autosomal dominant polycystic kidney disease in two other family members. The proband was homozygous for the "Maghrebian" mutation in the AGXT gene, confirming the diagnosis of primary hyperoxaluria type 1[1, 2]. The coexistence of these two diseases in the same family raises complex issues of genetic counseling and renal transplantation. The father remains the only potential living donor, but careful evaluation is necessary [10]. The development of cadaveric organ transplantation in Algeria remains the only hope for this family.

Conflict of Interest:

The authors declare no conflicts of interest.

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