Research Article

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A Mutation in the OFD1 Gene is Seen in Polycystic Kidney Disease that is Linked to Intracranial Hypertension

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Abstract

We describe a very unusual instance of type I polycystic kidney disease and choledochal cysts. Due to polycystic kidney disease, a 48-year-old woman was reliant on hemodialysis for chronic renal failure. By chance, an ultrasound revealed that she also had a dilated common bile duct. An endoscopic retrograde cholangiopancreatography without visualisation of the pancreatic duct revealed a spindle-shaped, dilated common bile duct (type I choledochal cyst). The choledochal cyst was removed from her. A cholangiogram performed during surgery revealed no evidence of contrast medium reflux into the pancreatic duct. The aspirated bile from the bile duct did not have an increased amylase level. The presence of renal fibro polycystic disease may prevent pancreaticobiliary dysfunction from becoming a factor in the aetiology of choledochal cysts. In these situations, choledochal cyst management is still debatable and needs more discussion.

Keywords: Kidney disease • Hypertension • Endoscopic • Pancreatic duct • Polycystic kidney disease • Endocrinological disorders

Introduction

Idiopathic intracranial hypertension (IIH), commonly referred to as pseudotumor cerebri, has an unknown aetiology. IIH typically affects young, obese women. Numerous studies have reported underlying and related endocrinological disorders. The presence of signs and symptoms of intracranial hypertension, such as papilledema, documented elevated intracranial pressure, and normal CSF composition, the absence of a mass, structural lesion, or vascular lesion on MRI, MR venography, or contrast-enhanced CT, and the absence of another intracranial hypertension-causing factor are all diagnostic criteria for IIH [1]. The main goals of treatment are to prevent visual impairment and to relieve headaches. It is advised to take acetazolamide, lose weight, and address underlying risk factors, such as endocrinological abnormalities. Some conditions are unresponsive to medical treatment and necessitate surgery (lumboperitoneal shunting or fenestration of the optic-nerve sheath). In order to prevent visual compromise, women with IIH require all the assistance we can provide to help them combat obesity and endocrinological disorders [2].

Truncal obesity, irregular menstruation, hyperandrogenism with hirsutism, acne, alopecia, and many ovarian follicular cysts are all symptoms of the polycystic ovary syndrome (PCOS). A number of endocrine anomalies, including an enhanced conversion of androstenedione to estrone by stromal cells in adipose tissue, are indicative of PCOS. In PCOS, insulin resistance is more prevalent. For defining PCOS, there are two major acknowledged approaches. Two requirements must be accomplished in order for the NIH criteria to be satisfied: persistent anovulation and clinical or biochemical proof of hyperandrogenism. The American Society for Reproductive Medicine (ASRM) and the European Society for Human Reproduction and Embryology (ESHRE) recently proposed revised criteria for the classification of PCOS [3].

It is a multisystem genetic disorder called tuberous sclerosis. Numerous cancers with involvement of the kidney and overexpression of the mTOR gene result in end-stage renal failure. When a patient

enters adulthood, renal polycystosis develops as a result of the growth of numerous cysts, angiomyolipomas, and carcinomas in the renal parenchyma. We present a 30-year-old man who has end-stage renal disease brought on by renal polycystosis and tuberous sclerosis. Renal function, tuberous sclerosis symptoms, and renal polycystosis all markedly improved after immunosuppressive therapy with rapamycin. In order to help identify women with IIH who are at risk for PCOS, we also recommend that neurologists and ophthalmologists use a screening questionnaire [4].

Because somatostatin analogues are cAMP inhibitors, they can lower fluid secretion and cholangiocyte proliferation. After 6 to 12 months of treatment with lanreotide, a substantial decrease in liver volume was achieved when compared to placebo, according to two randomised controlled trials. The abdominal symptoms did not dramatically improve, and the average liver volume reduction was just 3% to 5%. The risks of surgical intervention may not be warranted in this particular group of patients with symptomatic PLD, or it may be technically difficult to perform the surgery [5].

Mammalian target rapamycin (m-Tor) inhibitors, on the other hand, exhibit antiproliferative and immunosuppressive effects. However, while some animal model studies looked to have promising findings, other research failed to demonstrate significant therapeutic effects, whether neither in people nor in animal models. Percutaneous cyst aspiration, laparoscopic or open cyst fenestration, hepatic resection, and transplantation are the most popular interventional and surgical therapy options. Due to the huge size of the liver and displacement of common anatomic landmarks in these patients, resection and transplantation are technically challenging procedures. In an ADPKD patient with liver cysts, ascites and variceal haemorrhage are infrequent, but when they do occur, further testing to rule out vascular thrombosis is necessary. Huge liver cysts may result in a subsequent Budd-Chiari syndrome and a hepatic venous outflow blockage. The patient may be a candidate for a combined liver and kidney transplant in this situation and if they have ESRD.

This case study describes a 59-year-old lady who had end-stage renal disease and ADPKD who also acquired a secondary Budd-Chiari syndrome and was successfully treated with a liver and kidney transplant. After nine years of follow-up, the patient is still asymptomatic and has fully resumed everyday activities. This case highlights the potential for a double liver and kidney transplant to treat ADPKD patients who have liver cysts that have become aggravated by the emergence of a Budd-Chiari syndrome [6].

Discussion

In order to help neuro ophthalmologists identify women with IIH who may develop PCOS, we looked at the use of a new screening questionnaire. According to this survey, 38% of women with IIH were at risk for developing PCOS. In 15.5% of the study participants, additional blood tests and an endocrinologist examination aided in the diagnosis' clarification, the proposed screening tool's positive predictive value, which used a questionnaire to assess PCOS risk, was 41% (9/22). Additionally, we discovered that the IHH study group had a considerably greater frequency of PCOS (15.5%) as compared to the general population when utilising the NIH criteria. Based on a consensus of specialists, the NIH diagnostic criteria for the diagnosis of PCOS state that women have the condition if they exhibit chronic oligo- or anovulation in addition to biochemical or clinical symptoms of hyperandrogenism. Their theory, which contends that androgen excess, is a key aspect of the condition, served as the foundation for our questionnaire [7].

As was the case in earlier community studies of PCOS prevalence, ultrasound and blood tests were not done on women who did not exhibit clinical signs of PCOS. It's possible that some of the women in this group had polycystic ovaries and high androgenemia; although research indicates that this condition affects less than 1% of people with PCOS. We discovered that the IHH study group had a greater prevalence of PCOS using the NIH criteria (15.5%) compared to the general population (8.7%) when comparing the findings of the current investigation to earlier studies in the general population with the same age and BMI distribution. results were much lower in three additional earlier investigations that used the NIH criteria, with results ranging from 6.5 to 6.8% [8].

The findings of this investigation support a prior finding that IIH and PCOS are related. Reported that 15 (39%) of the 38 women with IIH who had PCOS. Another study indicated that 37 (57%) of the 65 women with IIH had PCOS. claimed that the prevalence of PCOS in women with IIH (39%–57%) is 5–8 times higher than the prevalence of PCOS in the overall unselected group (7%). These results serve as the foundation for this study's verification of the claim. The fact that Glueck used the less stringent Rotterdam criteria for PCOS rather than the NIH criteria may account for the disparity in prevalence. Although our study group is inconveniently small, it was found to be comparable in terms of clinical parameters when compared to one of the most comprehensive investigations of PCOS prevalence in Caucasian women to date [9].

For the first time, we advocate the use of a simple questionnaire that patients can complete on their own to help identify PCOS risk in a neurology or ophthalmology clinic setting. Since the patients can also be directly questioned in the event that the neuro ophthalmologist recalls the probable relationship between PCO and IHH, the questionnaire itself is not very important. Additionally, we did not formally validate this questionnaire. To emphasise the importance of detecting the associated disorders in order to enhance the management of IHH patients, we do offer this tool. By assisting with weight loss and managing accompanying endocrinological abnormalities such hyperandrogenism and glucose intolerance, treating PCOS may aid in the treatment of IIH [10].

Conclusions

Our clinical study demonstrates that rapamycin is a suitable substitute for an immunosuppressant in tuberous sclerosis patients getting a kidney graft. It displays the benefits compared to other immunosuppressive medications, including the improvement of renal cysts and the regression of tumoral lesions. Additionally, it would suggest a better pharmacokinetic profile in the concurrent use of inductive medicines, which are widely used to treat a variety of illness symptoms in the aforementioned patients. We come to the conclusion that rapamycin ought to be a first-line treatment for tuberous sclerosis patients.

Acknowledgement

None

Conflict of Interest

None

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