

Correspondence

Bilateral multicystic renal dysplasia with Potter sequence

To the Editor

I have read with interest the case report "Bilateral multicystic renal dysplasia with Potter sequence. A case with penile agenesis" by Dursun et al.¹ The authors stated that the case "provides most symptoms of bilateral multicystic hereditary renal adysplasia (HRA)". Although most cases of renal adysplasia (agenesis/dysplasia) are sporadic, some cases are inherited. Hereditary renal adysplasia is probably inherited as an autosomal dominant condition as the authors stated. Therefore, when renal adysplasia is diagnosed, careful screening (renal ultrasound) of the parents, sibling, subsequent pregnancies, and other relatives are important for the purpose of accurate genetic counseling,² as this anomaly can be asymptomatic. This was not performed in this family, and therefore one cannot state that this stillborn had an inherited disease. Although this child had a bilateral involvement and was a stillborn, this does not necessarily mean that affected family members should have a bilateral involvement and therefore will be symptomatic. Affected relatives of a proband with bilateral disease may have a unilateral involvement as this anomaly has variable expression,³ which supports the importance of screening in this family as the presence of a single normal kidney needs special precautions. Furthermore, the authors stated: "the penile agenesis was first reported and the consanguinity in the parents might further delineate the bilateral multicystic HRA". This statement may not be accurate as appropriate family screening was not performed and since it is known that the most usual mode of inheritance of this disease is through an autosomal dominant gene, which is not affected by consanguinity. The presence of consanguinity may be a coincidence. Moreover, absence of this anomaly in the parents does not rule out an autosomal dominant inheritance² as the gene may have incomplete penetrance; hence, aunts, uncles, and grandparents may have to be screened. Penile agenesis is not a new association with multicystic dysplastic kidneys as it was implied; an earlier report documented a similar association.⁴

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Reply from the Author

We greatly appreciate the comment from Dr. Fuad I. Abbag. First, we would like to state that the parents of the child had physical and laboratory examination for any anomaly that may present itself related to the present condition. Unfortunately, for insurance purposes, the family did not want us to further investigate the matter thoroughly (radiologically, ultrasound, and so forth). As our patient was the first child of the family, no siblings were present as mentioned in the manuscript. We also tried to reach the recent relatives of the family, however, they were distant and we only obtained the information that no symptomatic individuals were present. Unfortunately, due to space limitation as this is a report of a case, we did not extensively discuss this situation in our paper, however, we are thankful to now have this opportunity to do so. Secondly, as we all know, most of the cases are sporadic and the rest is suspected to be inherited by autosomal dominant trait. We agree to the criticism that our case might be a result of either. However, the presence of consanguinity had to be implied for a future reference. We have not intended to suggest that our case was inherited by an autosomal recessive trait. We accept the fact that incomplete expression of the gene, which is unknown and suggested to be a dominant in nature, from both parents might be the cause of the situation. Nonetheless, consanguinity and a remote possibility of the recessive trait had to be stated. Lastly, in our search, penile agenesis and consanguinity were not documented for this matter. We agree that this should have been clearly emphasized in the manuscript, and thanks to Dr. Fuad I. Abbag, we have an opportunity to share this with the readers.

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