Answer to Medical Quiz - 2

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Answer

1. Enlarged kidneys with multiple cysts, hepatic cysts
2. Autosomal dominant polycystic kidney disease (ADPKD)
3. Mitral valve prolapse
4. Rupture of intra-cranial aneurysm

Review

Autosomal dominant polycystic kidney disease (ADPKD) is the most common inherited kidney disease accounting for 1 in 800 live births in western world with 5-10% patients in end-stage renal disease (ESRD) programm.[1,2] Mutations may be in PKD-1 gene (located in chromosome 16, common and have relatively aggressive disease) and PKD-2 gene (located in chromosome 4). Defective synthesis of polycystin-1 and 2 results in cyst formation in kidneys and other organs. Patients may remain asymptomatic; common presentations include hypertension, abdominal heaviness, pain, haematuria, renal failure, etc.[3] Ultrasonography is the investigation of choice and also used for screening of first degree relatives over 20 years of age. Abdominal pain may result from cyst infection, expansion, haemorrhage and stone. Treatment is directed towards control of hypertension by angiotensine blocking agents, treatment of infection and monitoring. Newer agents may help reduce cell proliferation.[1] Patients with progressive kidney failure may require kidney transplantation. As the disease has an autosomal dominant inheritence, each of the off spring has a 50% chance of being affected and this issue should be discussied during counselling.

References

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