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Variable clinical features in patients with Fabry disease

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We report our experience with Fabry disease in a multidisciplinary clinic in UC Irvine. Our cohort comprises two children and 17 adult patients (10 M and 9 F) ranging in age from 6- 62 y., with age of diagnosis ranging from 3-57. The data shows that 42% of all patients suffer gastrointestinal problems, 53% have reduced sweating, 63% have angiokeratomas, 58% exhibit corneal whorls, 32% have lymphedema, and 90% of the male patients experienced acroparesthesia. 53% of adults have tinnitus, 41% of adults experience hearing loss. There is renal involvement in 37% of patients, most have varying levels of proteinuria but one required a kidney transplant. Thirty-two percent of patients have evidence of cardiomyopathy determined by echocardiogram and MRI studies. One male had a stroke and currently has MRI changes of multifocal encephalomalacia and adjacent gliosis of the left cerebral hemisphere, frontal and parietal lobes. Ninety percent of the males, 44% of the females, and 50% of the children currently receive enzyme replacement therapy (ERT). Each individual demonstrates different response rates to symptom improvement and slowing of organ deterioration. Two individuals developed infusion reactions, one male developed ERT associated meningitis in 2009 which prompted him to end treatment. He re-initiated ERT May 2015 and has continued to tolerate ERT with premedication. ERT will be initiated in the 6 year old male for reduced GFR, a urine GL-3 level of 569μg/mmol Cr. Clinical features are phenotypically heterogeneous, perhaps due to the variety of unique mutations in the GLA gene. Monitoring patients regularly provides insight into genotype- phenotype correlations thus leading to optimization of patient care.

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