Primary hyperoxaluria Type 1: indications for screening and guidance for diagnosis and treatment.

Abstract:
Primary hyperoxaluria Type 1 is a rare autosomal recessive inborn error of glyoxylate metabolism, caused by a deficiency of the liver-specific enzyme alanine:glyoxylate aminotransferase. The disorder results in overproduction and excessive urinary excretion of oxalate, causing recurrent urolithiasis and nephrocalcinosis. As glomerular filtration rate declines due to progressive renal involvement, oxalate accumulates leading to systemic oxalosis. The diagnosis is based on clinical and sonographic findings, urine oxalate assessment, enzymology and/or DNA analysis. Early initiation of conservative treatment (high fluid intake, pyridoxine, inhibitors of calcium oxalate crystallization) aims at maintaining renal function. In chronic kidney disease Stages 4 and 5, the best outcomes to date were achieved.
with combined liver-kidney transplantation.

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