Comment on Gitelman's syndrome: Rare presentation with growth retardation

Sir,

We read with interest the article "Gitelman's syndrome (GS): Rare presentation with growth retardation".[1]

The authors say that presentation of GS with growth retardation is rare. They also state that they have not found any reported cases of GS in childhood and that theirs is probably the first ever case of GS to be reported in a child. However, neither of these claims is tenable since GS has been described in children and growth retardation is a common presenting complaint when the onset is early.

Though GS has classically been considered to be a benign condition with onset in adolescence or childhood with patients being asymptomatic or having mild symptoms, clinical studies have revealed that patients with GS may show considerable phenotypic variability.

Schmidt *et al*., described the clinical, biochemical and molecular genetic data in 5 children with GS.[2] Riveira-Munoz *et al*., in 2007 described mutations associated with a sub-group of patients presenting with "severe" symptomatology such as early onset, growth retardation severe neuromuscular manifestations such as tetany and seizures, rhabdomyolysis, chondrocalcinosis and ventricular arrhythmia. In fact, their study suggested that the nature/position of the *SLC12A3* mutation, combined with male gender, is a determinant factor in the severity of GS. In their series, they had 6 children with onset at 12 years of age or less, with 2 boys being 21 months and 18 months at presentation.[3]

Tammaro *et al*. described GS in two sets of prematurely born twins who presented with hypokalemia in the 1st month of life while Skalova *et al*., described an 18-month with GS who presented with psychomotor retardation and failure to thrive.[4,5] Sinha *et al*., from New Delhi also described a child with GS with onset at 2.5 years of age.[6]

It is important to do an extensive literature search using Medical Subject Headings (MESH terms) to avoid missing relevant references. It might be worthwhile doing mutational analysis in this child due to the early age of presentation.

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