

SYSTEMIC MANAGEMENT

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on behalf of Detlef Bockenhauer and Lorenzo Calo

Disclosure of Interests

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Joints

Chondrocalcinosis and pseudogout are both common

- Calcium pyrophosphate crystals
- Mg required for pyrophosphatase activity
- Case reports, case series and one cross-sectional study
- Prevalence unknown, severity variable
- May require joint replacement

Increase Mg

? NSAIDs, colchicine



Growth

Epidemiology of growth impairment unknown

- Few case reports and one series only; conflicting evidence
- GH deficiency recognized in Bartter syndrome
- One report of electrolyte supplementation helping

? Assess for growth hormone deficiency if electrolytes well supported and growth still sub-normal?



Two small studiesa) 10/16 Chinese had impaired toleranceb) 0/5 Caucasian (indeed insulin sensitivity up)

- Based on GS's similarity to thiazide Rx
- However unknown if this is on- or off-target

? Monitor HbA1C



Cardiovascular management and sport

Hypokalemia associated with prolonged QT interval

Evidence of LV dysfunction with reduction of myocardial perfusion, myocardial contractile recruitment and cardiac index

However, correlation with low K/Mg unclear

Might be precipitated by extreme exercise?

Anecdotal reports of sudden death

? EKG esp focusing on QT

? Avoid extreme sport

Avoid drugs that prolong QT interval



Pregnancy

Increased GFR, aldo and volume will worsen electrolyte disturbances

- Vomiting may be problematic
- Some Rxs contraindicated
- HypoMg associated with increased fetal loss in rodents
- ? Additional risks during labour

Increase supplements including IV where necessary Stop ACEI/ARB; amiloride probably OK. ?spiro/eplerenone Frequent monitoring during labour



Perioperative management

One case series and other literature on hypoK

- Prolonged neuromuscular blockade
- N/G suction and hyperventialation
- Adenaline

Frequent monitoring

IV replacements

Resume oral Rx asap

UK has NICE-approved guideines



Patient education and support

'Patient in the driving seat'

- Keeps blood results
- Knows pathophysiology
- Understands compliance and risks
- Dietary education

This information can be provided through a variety of media Personal tuition in a clinical setting Patient information leaflets Web-based information Patient-led fora (eg <http://www.gitelmansyndrome.co.uk/>) Patient and family groups' support events



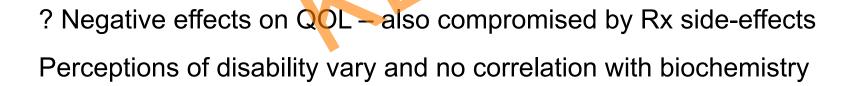
Patient education and support

No rules for follow-up frequency

? ID bracelet

Doctor's letter for travel

Occupational health (eg nightshifts)



Sick day rules



If you are ill, do not stop taking your supplements unless advised by your doctor

If you have diarrhea or vomiting, increase your fluid intake but ensure that you add salt, or some electrolyte powder, to anything you drink. Also stop ACE-inhibitors/ARBs until you can keep fluids down If you are vomiting and cannot keep anything down for 24 h or more, you should seek immediate medical advice Seek immediate medical advice if you:

become dizzy

develop tingling or muscle weakness

notice an irregular heartbeat (palpitations)

See your doctor if you notice any unusual symptoms, as your medication dosage might need altering; a simple blood test might be all that is needed



Not written yet / for further discussion

What biochemical analyses should be undertaken to look for complications in GS patient? (e.g.,lipid, creatinine, proteinuria testing, etc.)

• 6/36 adults elevated ACR (Berry et al 2012)

Also hypertension

?- 44% in reports

Is there a utility in developing a clinical score based on the multisystemic manifestations? Probably not – variable severity, multiple symptoms, utility not obvious.

(Inter)national registries



Longterm mx eg cysts

KDIGO Controversies Conference on Gitelman Syndrome | February 12-13, 2016 | Brussels, Belgium