SYSTEMIC MANAGEMENT

Fiona Karet
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?

Glucose intolerance?

Two small studies

a) 10/16 Chinese had impaired tolerance

b) 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)

? Negative effects on QOL – also compromised by Rx side-effects

Perceptions of disability vary and no correlation with biochemistry

Sick day rules

‘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 multi-systemic manifestations? Probably not – variable severity, multiple symptoms, utility not obvious.

- (Inter)national registries
- Vitamin D

Longterm mx eg cysts