Kleefstra Syndrome is a rare genetic disorder, caused by a EHMT-1 gene deletion or mutation. Most kids with Kleefstra Syndrome have some physiological and behavioural/psychiatric/sleep differences that make it difficult to assess how sick they actually are.

Top tips for triaging & treating kids with Kleefstra Syndrome

1. Poor temp control:

May not develop a fever, or hypothermia may be normal for them (and so "normothermia" would be pyrexial for them). May not sweat at all.

2. Weak immune system:

Poor response to vaccinations; atypical presentations of infections. Pneumonia is common and often associated with swallowing difficulties (see #8)

3. Beware fractures

People with Kleefstra Syndrome are stoical, and have been known to walk on fractures (including femurs!) Osteoporosis is common, due to immobility and drug side effects.

4. Epilepsy is common

One-third of kids with Kleefstra Syndrome have epilepsy, and Sudden Unexplained Death in Epilepsy (SUDEP) common.

5. Beware the poop!

"Soft stool" constipation is common (you won't be able to feel it: this may be one of the few instances where using AXR to detect constipation is justified). Some people with KS need caecostomies or even colectomies to manage this.

6. Delayed gastric emptying

Vomiting/reflux is common. Erythromycin may be used long-term to increase gastric motility: watch out for drug interactions.

7. Beware the co-morbidities

People with Kleefstra syndrome are at risk of cardiac, renal, GORD, neurological (including hypotonia), central and obstructive apnoea, dental & ENT problems. Watch out for these, and take a careful drug history.

8. Swallowing problems

Very common: may be complicated by aspiration pneumonia. Tube feeding is common.

9. Ask what's normal for this individual child

Assessing alertness, tone, responsiveness etc. can be difficult if you don't know the individual child, so ask the parents! Also, be mindful that children with this condition typically understand a lot more than they can communicate.

10. The scary sleep thing

At or after puberty, severe sleep disturbance lasting >48 hours may signify psychosis and onset of regression (expect seriously worried parents). This is a same or next-day CAMHS emergency, requiring treatment with anti-psychotics. NB: beware paradoxical response to benzodiazepines!

Kleefstra Syndrome was only recognised in 2012, but there's a thriving community of parents whose children have this condition. They'll probably know more than you do, and also know from whom you should be seeking advice. Keep calm, and utilise parental power!

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