Methods We present case reports from a UK hospital focussing on 2 patients with rare genetic conditions associated with short stature- Trichorhinophalangeal Syndrome and KBG Syndrome, accompanied by a summary of the current literature surrounding rHGH use in each condition.

Results There are 10 reported cases of rHGH use in TRPS, summarised below.

<table>
<thead>
<tr>
<th>Paper</th>
<th>GH dose (mg/kg/week)</th>
<th>Height standard deviation score change</th>
</tr>
</thead>
<tbody>
<tr>
<td>Naselli 1998</td>
<td>0.23</td>
<td>0</td>
</tr>
<tr>
<td>Stagi 2008</td>
<td>0.26</td>
<td>+0.7 in 5 years</td>
</tr>
<tr>
<td>Sarafoglou 2010</td>
<td>0.3–0.43</td>
<td>+1.81 in 3 years</td>
</tr>
<tr>
<td>Sohn 2012</td>
<td>0.2</td>
<td>+0.4 in 10 years</td>
</tr>
<tr>
<td>Merjaneh 2014</td>
<td>0.28</td>
<td>+1.0 in 2 years</td>
</tr>
<tr>
<td>Riedl 2004</td>
<td>0.2</td>
<td>?</td>
</tr>
</tbody>
</table>

Taken together, they suggest rHGH can be of benefit, and that earlier initiation of therapy is associated with better height outcomes. More information is needed before TRPS I can be considered a firm indication, but rHGH has potential to improve height outcomes in the short term.

Turning attention to KBG syndrome, there are only 2 detailed case reports of GH treatment in this condition (Reynart et al. 2015). These children increased their height by 0.6 and 1 SDS within 1 year of treatment, respectively.

Conclusion This work demonstrates the clinical features of 2 rare genetic conditions, and highlights the need for further debate around the potential of rHGH in maximising growth potential, with the ultimate aim of improving quality of life for patients with rare conditions including KBG syndrome and TRPS.

REFERENCES


G233(P) CRANIOSYNOSTOSIS CAN OCCUR IN CHILDREN WITH NUTRITIONAL RICKET

Background Severe vitamin D deficiency (VDD) is a common disorder which has complications including rickets, hypocalcaemia, hypotonia, delayed development and cardiomyopathy. Although nutritional rickets associated craniosynostosis has been reported, there is little awareness of this or knowledge about its clinical course or severity. We present five cases of late onset craniosynostosis in association with nutritional rickets.

Clinical presentation The diagnosis of craniosynostosis was made between the age of 16 months and 3 years (n=5). All children had clinically evident scaphocephaly and radiological evidence of previous rickets. All children had risk factors for severe VDD: African or Asian backgrounds with darker skin pigmentation (n=5); multiple food intolerances (n=2) and prolonged breastfeeding with picky eating habits (n=2). They presented in two ways:

Group 1 (n=3) presented with clinical and radiological signs of severe rickets after a long period of untreated severe VDD. Serum 25OH vitamin D levels<20 nmol/L, elevated alkaline phosphatase, elevated parathyroid hormone (PTH) concentrations, low serum calcium and low phosphate concentrations. They were managed with treatment doses of vitamin D and calcium supplementation where necessary. In two patients, treatment had been completed and clinical signs resolved when the craniosynostosis was diagnosed.

Group 2 (n=2) presented with sagittal suture ridging and scaphocephaly associated with resolving rickets on radiology. Clinically there were other signs of VDD and serum 25OH vitamin D concentrations were 33–44 nmol/L with normal PTH and bone profiles.

CT in all cases showed fusion of the sagittal sutures. Three of the children also had multiple suture fusion. All in Group 1 were managed conservatively but Group 2 patients had raised intracranial pressure and both underwent surgical cranial vault remodelling.

Conclusions All the patients had nutritional rickets associated with craniosynostosis. Patients with late presentation and sagittal suture ridging went on to have emergency cranial vault remodelling. It is important to recognise this complication early and refer to the neurosurgeons and so prevent raised intracranial pressure. It is important to collect detailed data on this and study a larger cohort to raise awareness, establish the pathophysiology and try to prevent this complication.