Complex Gastrointestinal (GI) Issues in CHARGE Syndrome

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Background

- CHARGE syndrome is a complex genetic syndrome often involving midline structures, with limited research on the gastrointestinal (GI) system.
- A zebrafish model of CHARGE Syndrome has demonstrated dysmotility and there is data to support abnormalities in the microbiome[1].
- Those with cranial facial abnormalities and a history of gastrointestinal tube feeding have been shown to have significantly more GI symptoms[2].

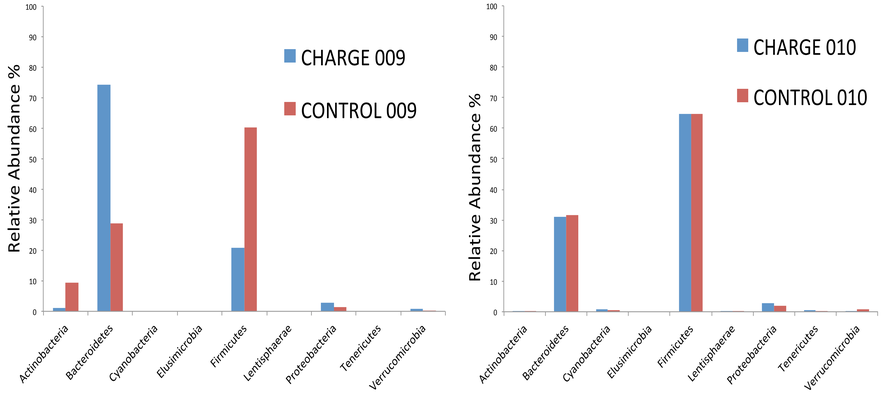


Figure 1. Relative abundances of gut microbes in a CHARGE Syndrome sibling pair with different GI scores (left) and similar GI scores (right).

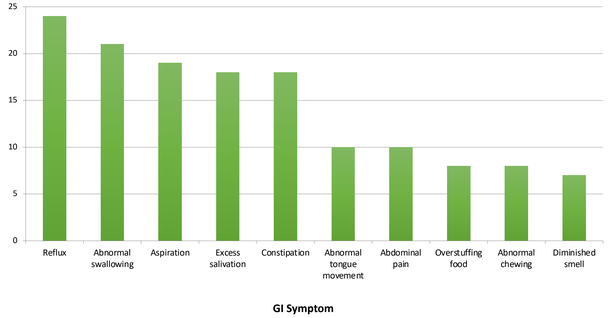


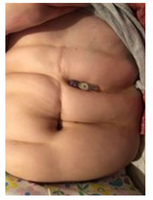
Figure 2. Absolute frequency of parent reported GI symptoms in children with CHARGE Syndrome[2].

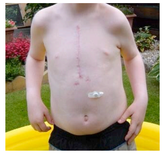
5 year old with regression of oral intake following acute illness and psychosocial stressors

- Initially the child was successfully eating a puree diet by mouth with minimal GI complaints with SLP support at school.
- With the COVID-19 pandemic shutdown and subsequent contraction of COVID-19, the child had new reflux, constipation, abdominal pain and refusal to eat by mouth.
- No improvement with classic reflux or constipation medications.
- Symptoms only improved with return to routine and increasing sensory enjoyment of foods with varying temperatures and textures.

32 year old with recurrent bowel obstruction due to congenital volvulus

- During the third presentation of decreased oral intake and feculent emesis, indicative of a bowel obstruction, the decision was made to perform an exploratory laparotomy.
- Due to choanal atresia an NG tube was unable to be placed for decompression.
- The obstruction was secondary to a partial congenital volvulus.
- There were incidental findings of a floppy cecum and minimal retroperitoneal attachments of the right colon.
- After surgery, the individual's mood was significantly improved, and for the first time in their life was able to lay on their right side and was agreeable to wear their cochlear implant.





32 year old with severe dysmotility and bowel obstruction

- Despite partial congenital bowel volvulus repair there have been recurrent bowel obstructions secondary to constipation resulting from severe dysmotility.
- All testing revealed normal GI anatomy.
- More than 15 medications and natural supplements, and a liquid diet have been tried with no improvement in abdominal pain or constipation.
- Next steps include TPN or bowel pacer for symptom relief.

5 year old with cyclic abdominal pain and vomiting due to migraines

- Abdominal pain and vomiting occurred every 1-2 months for 3-6 days.
- All GI investigations were normal and classic treatments for constipation and nausea lead to more vomiting.
- There was a family history of migraines, but the individual was unable to communicate migraine symptoms.
- A symptom diary revealed triggers of sleep deprivation, stress and dehydration. Symptoms improved with avoidance of triggers.

10 year old with severe reflux resulting in recurrent respiratory arrest

- Feeding intolerance and severe reflux started from birth and resulted in recurrent episodes of respiratory distress and apnea.
- The reflux and feeding intolerance was refractory to medications.
- Ultimately long-term home TPN was initiated due to risk of further damage to the respiratory system with repeated aspirations.

Conclusions

Complex GI issues in CHARGE Syndrome are not uncommon and are likely due to a combination of dysmotility, abnormalities of the microbiome and anatomical abnormalities. Even common conditions such as chronic constipation can be difficult to manage and cause distress to both patients and families. Early involvement and collaboration of a multidisciplinary team and validated tools like the feeding scale for individuals with genetic syndromes may benefit those with CHARGE Syndrome[2].

References

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Disclosures

I do not have any conflicts of interest to disclose.