Nutritional Support Is Required in the Majority of Patients with CHARGE Syndrome: 10-year Data from a Single Tertiary Centre

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Introduction

CHARGE syndrome is a rare genetic disorder, caused in more than half of cases by mutations in the CHD7 gene. CHARGE is an acronym for the clinical features common in this condition- Coloboma of the eye, Heart defects, Atresia of the choanae, Restriction of growth and development, Genito-urinary abnormalities, and Ear abnormalities and deafness.

Gastrointestinal (GI) symptoms, feeding difficulties and growth restriction are known to be highly prevalent in patients with CHARGE syndrome. We describe our experience with children diagnosed with CHARGE syndrome over a 10-year period in our tertiary paediatric gastroenterology unit.

Subjects and Methods

We reviewed case notes of patients with CHARGE syndrome managed in our hospital from 1 January 2010 to 31 July 2021. Patients were identified based on a diagnosis of CHARGE syndrome by genetic or clinical criteria.

Results

Sixteen patients were included in the study. 11 patients (69%) were female. No deaths were recorded. Median age of patients at the time of review was 10.3 years (range 0.9- 15.8 years). Eleven patients had confirmed CHD7 gene mutations, the remainder were diagnosed clinically.
Clinical features in our patient cohort are described in Table 1.

Most patients had feeding difficulties, with 14 patients (88%) requiring a period of enteral feeding support in the form of gastric feeding (n=14) and also jejunal feeding (n=4). One patient required five months of parenteral nutrition due to vomiting, recurrent aspiration events on feeds and faltering growth. At last review, six patients had been completely established on oral feeds, while the remainder still required enteral feeding support.

Gastro-oesophageal reflux disease (GORD) was diagnosed in 11/16 patients (69%). Of these, seven patients underwent pH/impedance study; three patients had evidence of GORD while the remainder had normal studies. Three patients underwent upper gastrointestinal endoscopy; all had normal results.

In terms of surgical interventions, ten patients (63%) had percutaneous endoscopic gastrostomy (PEG) insertion; two patients also underwent fundoplication. One patient had a percutaneous endoscopic gastrostomy-jejunostomy (PEG-J) insertion and one had a Roux-en-Y jejunostomy. One patient underwent oesophageal gastric disconnection surgery at the age of two years due to persistent retching and vomiting. Post-surgery, vomiting improved however the patient continued to retch.

Clinical Feature	Frequency in Cohort (%)
Gastrointestinal	
Feeding difficulties	14/16 (88%)
Gastro-oesophageal reflux disease	11/16 (69%)
Constipation	3/16 (19%)
Unsafe swallow	8/16 (50%)
Other Features	
Choanal atresia	7/16 (44%)
Coloboma	13/16 (81%)
Hearing defects	16/16 (100%)
Cardiac anomalies	14/16 (88%)
Endocrine issues	7/16 (30%)
Developmental delay	7/16 (44%)
Requirement for Tracheostomy ventilation	5/16 (31%)

Table 1: Clinical feature in our patient cohort

Conclusions

CHARGE syndrome can vary widely in severity; our cohort represents the more severely affected patients requiring intensive tertiary care input. Our study highlights the complexity of care and importance of multi-disciplinary input for these patients. Feeding issues are common in patients with CHARGE syndrome and many patients with CHARGE syndrome will require long-term enteral feeding support. Individual treatment plans should include an ongoing evaluation of gastrointestinal and feeding problems as part of the standard of care, to reduce morbidity and improve quality of life .

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