ORIGINAL ARTICLE

Treatment of hypophosphataemic rickets in children remains a challenge

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INTRODUCTION

Hypophosphataemic rickets (HR) is a rare hereditary disease characterised by hypophosphataemia, defects in bone mineralisation and rickets.

MATERIAL AND METHODS

We searched the hospital files at H.C. Andersen Children's Hospital, Odense University Hospital, Denmark, for children with the International Classification of Diseases 10 codes E83.3B (vitamin D resistant rickets) and E83.3A1 (familiar hypophosphataemia) from 1 February 2012 to 1 May 2012. Data were collected retrospectively.

RESULTS

Fifteen HR children were identified. X-linked hypophosphataemia with mutations in the phosphate-regulating endopeptidase homologue, X-linked were present in 80%; three had autosomal recessive HR with dentin matrix protein mutations. The children were treated with phosphate and alphacalcidol for an average of 7.7 years ± 5.1 standard deviations (SD). At the latest follow-up, the mean age was 10.1 (± 5.4) years, and the mean height had declined 0.8 SD from the first contact. A total of 40% had an actual height below –2.0 SD, and 40% underwent surgery for leg deformities. Among the medically treated patients, five had genu varus with a mean medial femoral condyle distance of 6.6 cm (± 2.79), and two patients had genu valgus with a mean medial malleolus distance of 12.3 cm (± 1.77). Episodes of secondary hyperparathyroidism were seen in 87%, and one patient developed transient nephrocalcinosis.

CONCLUSION

The current medical treatment for HR is insufficient. The rarity of the disease and the treatment difficulties of HR call for centralised management. International multi-centre trials including novel treatment options are warranted.

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SYSTEMATIC REVIEW

Single port laparoscopic rectal surgery – a systematic review

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INTRODUCTION

Single-port laparoscopic surgery (SPLS) for colonic disease has been widely described, whereas data for SPLS rectal resection is sparse. This review aimed to evaluate the feasibility, safety, and complication profile of SPLS for rectal diseases.

METHODS

A systematic literature search of PubMed and Embase was performed in September 2013 according to the PRISMA guidelines. Original reports on the use of SPLS in high and low anterior resection, Hartmann's operation and abdominoperineal resection were included. Outcome measures were intra-operative details and complications, short-term oncological outcome, and early complication profile.

RESULTS

No randomized studies or controlled clinical studies were identified. All studies were case series or case reports. Only five studies included more than ten patients operated with SPLS, comprising a total of 120 patients. These studies formed the basis for the final analyses on outcome. Operative times were 79-280 min. Conversion rates to conventional laparoscopic surgery and to open surgery were 12% and 2.5%, respectively. Numbers of harvested lymph nodes in malignant cases were 13-18. Post-operative complication rates were 25.5%. Length of hospital stay was 1-16 days. No 30-day mortality was reported.

CONCLUSION

Short-term results suggest that SPLS for rectal disease is feasible and safe with acceptable complication rate when performed by experienced surgeons in selected patients. Oncological safety and the possible benefits remain to be proven. Future rectal SPLS procedures should be done in a protocolled set up.

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