Intestinal ganglioneuromatosis as first manifestation of multiple endocrine neoplasia 2B in a premature girl

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INTRODUCTION

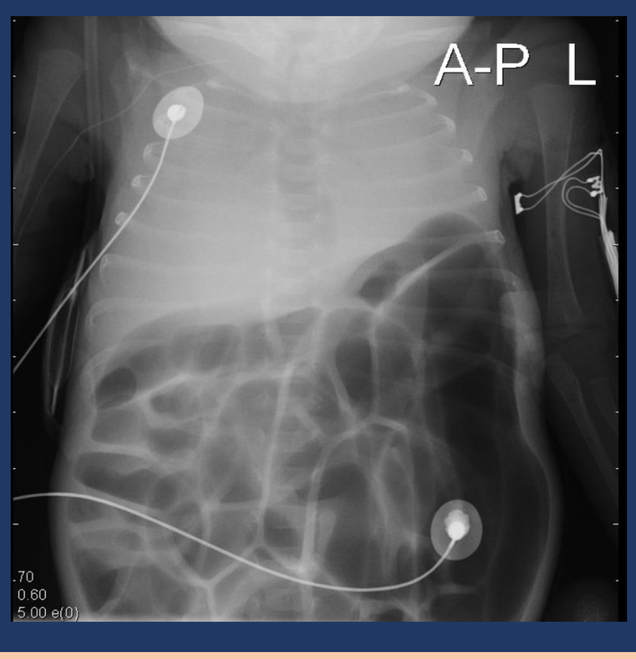
MEN2B is a rare cancerous syndrome caused by germline mutations in the *RE*arranged during *T*ransfection (*RET)* proto-oncogene. Inheritance is autosomal dominant however > 80% of cases are de novo mutations in M918T (95%) and A883F (<5%) within the *RET* gene.

Patients develop mucosal neuromas, corneal hypertrophy, musculoskeletal symptoms, severe constipation and a marfanoid body habitus.

Diagnosis on clinical features is suboptimal and often late as aggressive thyroid cancer before age 1 year is a hallmark of this condition.

CASE

A girl born at gestational age 24 + 6 weeks presented from day 1 with aspirates, abdominal distention and constipation. Meconium passing occurred on day 3. Increasing amounts of green aspirates and lactate rise was observed leading to a laparotomy at age 3 weeks. Chronic paralytic ileus in accordance with dilated bowels on previous abdominal films, without NEC, malrotation or stenosis was found.



The girl was failing to thrive and had secondary compromised breathing. At 10 weeks she was given an ileostomy to deflate the bowels. The bowels were extremely dilated proximal to the stoma as opposed to distally where they were collapsed.

Intestinal ganglioneuromatosis was described on a specimen of excised bowel.

Genetic analysis confirmed the RET M918T germline mutation when the girl was 12 weeks old. This was aided by the maternal history as she carried this mutation.

DISCUSSION

Gastrointestinal problems with severe constipation and pseudo obstruction are common (65%) in MEN2B. Abnormal proliferation of intramural ganglion cells resembling symptoms of Hirschsprung' s disease is causative. As in our case this may present already in premature neonates.

Early onset and aggressive thyroid medullary carcinoma always occurs before age 1 year in MEN2B. Thyroidectomy before and after 1 year of age had cure rates of 83% and 15% respectively.

The high frequency of de novo mutations and unspecific symptoms in infancy challenges the recommendation of thyroidectomy before age 1 year.

CONCLUSION

Extra-endocrine features of MEN2B occurring in early life are important for clinicians to be aware of to suspect this condition. Early diagnosis and timely prophylactic thyroidectomy is paramount.

CONTACT and REFERENCES

References:
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