An Unusual case of Gastrointestinal Bleeding: Expecting the Unexpected

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Introduction

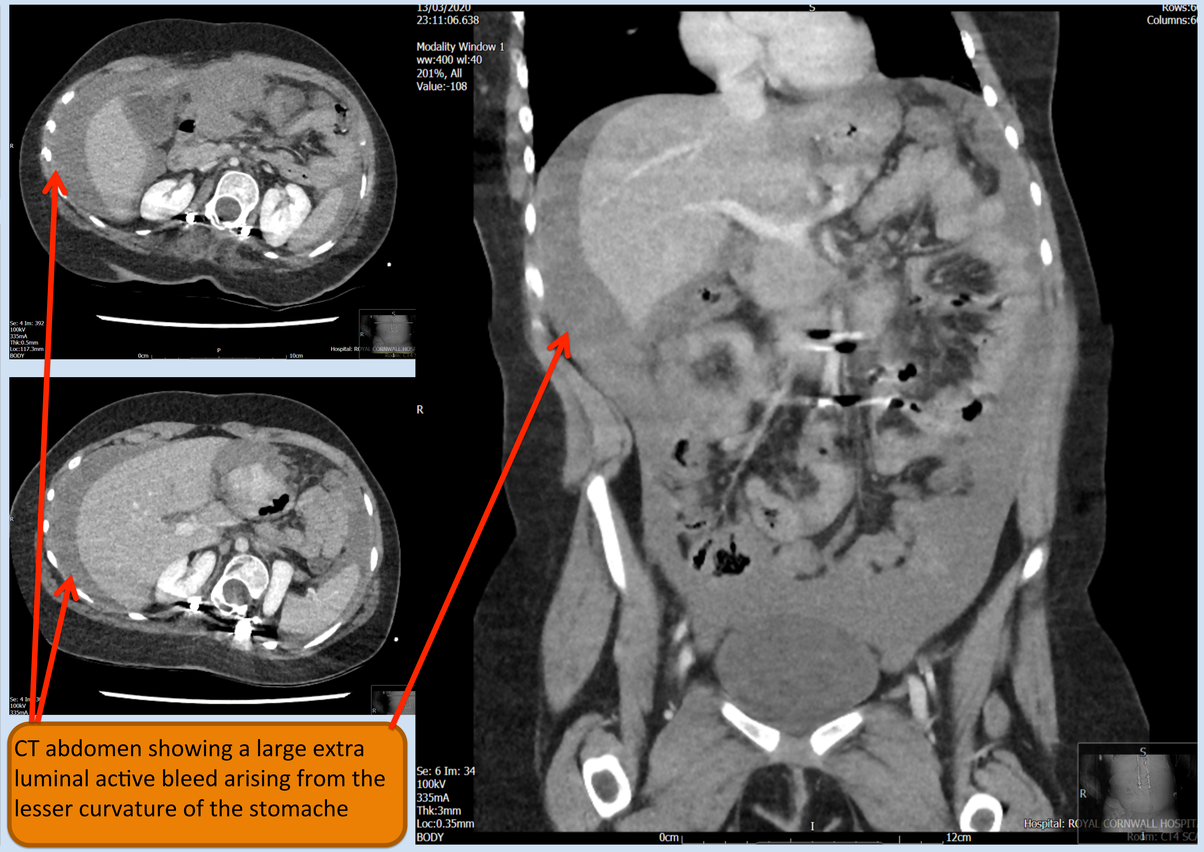
There is a known link between RASopathies such as Turners syndrome (TS) and solid tumours (gonadoblastoma, neuroblastoma and brain tumours,) [1] however no known link between TS and Gastrointestinal stromal tumours (GIST). The aim is to discuss a case of Turners Syndrome and GIST tumour in a single case study.

Case

An 11-year-old girl with background of Turner syndrome presented with large volume hematemesis, upper abdominal and shoulder tip pain, alongside clinical signs of shock. Past medical history included TS, cerebral palsy, autoimmune hypothyroid and surgically corrected scoliosis. Regular medication of oestrogen replacement, supra-physiological dose growth hormone therapy and levothyroxine. Blood results showed a metabolic acidosis, raised lactate, acute kidney injury and anaemia.
She required 40ml/kg fluid bolus and covered for suspected sepsis with ceftriaxone.

CT abdomen with contrast showed a large extra luminal active bleed from the stomach causing massive haemorrhage. Emergency laparotomy revealed the bleeding focus arising from a ruptured 7cm exophytic tumour in the lesser curvature of the stomach that was completely excised with clear margins. Later histopathology confirmed this to be a Gastrointestinal Stromal Tumour (GIST).

She was monitored and remained clear for reoccurrence, her growth hormone therapy was stopped due to a small increased risk of certain malignancies with a supra-physiological dose.



CT abdomen showing a large extra luminal active bleed arising from the lesser curvature of the stomache

Discussion

Gastrointestinal Stromal Tumours in children are mesenchymal in origin and rare, typically presenting in the second decade with a female predominance. They can occur anywhere in the gastrointestinal (GI) tract with the stomach and small intestine being the most common sites of disease [2]. Presentation is with anaemia from chronic GI blood loss, however acute haematemesis and melena is an unusual presentation in children and mandates the consideration for rare causes such as GIST tumours. There is differing histopathology between paediatric and adult GIST's with the paediatric form typically being benign. In this instance tumour molecular analysis revealed the most common paediatric histopathology of a Wild type GIST. Growth hormone (GH) therapy is recommended for Turners syndrome if proven GH deficient. Large-scale studies have shown an association between GH therapy and secondary malignancies on GH supra-physiological doses. [3]

This case presents insight into a rare paediatric tumour in a patient with known genetic condition Turners syndrome. There is an increased risk of certain solid tumours in Turners syndrome with no documented risk associated with rarer GISTs; it is an interesting case to document for future incidence.

References

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