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PARAGANGLIOMA AND GASTROINTESTINAL STROMAL TUMOR AS A SECOND MALIGNANCY IN A 14-YEAR-OLD GIRL; CARNEY TRIAD, CARNEY-STRATAKIS SYNDROME OR A NEW ENTITY?

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Carney triad is a rare clinical entity defined as a co-occurrence at least two of three following lesions: paraganglioma, gastrointestinal stromal tumor (GIST) and pulmonary chondroma. It is distinct from Carney – Stratakis syndrome: a familial condition of paraganglioma and GIST. To our best knowledge none of these have been reported as a second malignancy. A 16-year-old girl at the age of 3 had undergone neurosurgery followed by posterior fossa irradiation and chemotherapy due to cerebellar medulloblastoma. From that time she remains in remission with no visible CNS tumor and almost non-significant neurological sequel. At the age of 14 she was referred to our institution due to deep anemia and abdominal pain. There were neither vomiting nor weight loss. Abdominal ultrasound followed by gastrofiberoscopy revealed a large (7 x 4 x 3 x cm) tumor, infiltrating gastric wall, which turned out histologically to be GIST with marked C-kit (CD117) expression. Both the size and site of tumor were considered as contraindication for surgery, so she was given imatinib at the dose of 400, than 800 mg per day. Chest x-ray and subsequent chest CT showed highly vascularized mediastinal tumor. An attempt to dissect the tumor resulted in massive, almost fatal intraoperative bleeding, but the small tissue sample that could be obtained enabled the diagnosis of paraganglioma. Moreover this steadily growing mediastinal tumor started to compress airways, resulting in a respiratory distress. It did not respond to gemcitabine and docetaxel therapy (this was complicated by nephrotoxicity and subsequently withdrawn) however, when chemotherapy comprising cyclophosphamide, vincristine and dacarbazine (CVD) was administered, a partial tumor regression could be seen. The girl is well two years from diagnosis with the stable disease that seem to be controlled by administration of daily imatinib and cyclic CVD chemotherapy. A second surgical attempt is planned to remove both tumors. In contrast to Carney triad our patient's major medical problem is paraganglioma, however a negative family history speaks against the diagnosis of Carney-Stratakis syndrome. Since precise genetic studies are still unavailable we may only speculate on a genetic predisposition to a variety of tumors and a causative role of chemotherapy in developing concomitant GIST and paraganglioma.