GCT-P11 Genomic characterization of paediatric germ cell tumours

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Background: Germ cell tumours (GCTs) are characterized by distinct clinical and histological findings that influence the prognosis. For this reason, it is difficult to generalize the behavior of these tumours. GCTs have a low mutational rate and the most frequently altered genes are KIT, KRAS, and NRAS. Studies have shown that mutations in TERT or BRAF are rare or non-existent. However, most of these studies have been undertaken in adult patients and little is known about the molecular alterations that occur in paediatric GCTs. Our aim is to characterize the profile of genomic alterations in gene coding regions (exomes) in Brazilian paediatric patients with GCTs.
Methods: The exome analysis was performed for seven GCT samples, including two testicular, four ovarian, and one mediastinal using Illumina HiSeq 2500™ System. We reported the exome using Illumina paired-end sequencing strategy (>30 X-fold coverage) of seven cases and respective matched normal. Data analysis was performed as follows: (i) Mutect and Cancer Genome Interpreter; (ii) Nexus copy number software.
Results: Our data have shown that the most common genomic alterations found in paediatric GCTs were in high-risk patients that included gains in chromosome 12p. Among the classic genes involved in cancer biology, we found KRAS, PIK3C2G, MAP3K1, and ERBB3. The most frequently altered genes in GCT was CCND2, AURKA, AR, ERBB2, FANCA, FBXW7, FGFR1, INPP4BJUN, MYC, NKX2-1, PTEN, SUFU, and TNFAIP3. We identified that GCTS in a Brazilian paediatric population exhibit similar mutation profiles to those reported in other populations.

GCT-P12 Mediastinal germ cell tumours in children and adolescents: Report from the Polish Pediatric Solid Tumor Group

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Background: To present the biological and clinical features and management of mediastinal GCT (mGCT) in children and adolescents, based on the review of publications. We also analyse the patients with mGCT in Poland.
Methods: The publications from Medline were reviewed and patients with malignant mGCT and immature teratoma from Polish database from 2008 to 2018 were analysed.
Results: The published studies indicate some important differences in genetic profile, histology, presentation and outcome. The treatment results are worse than in other sites (EFS varies from 86% up to 100% in completely resected tumours). Among 362 patients with GCT registered in Poland, there were 11 (3%) with the mediastinal localization. Age: 7 patients <10 y and 4 patients >10 y. GCT markers were initially examined in 6 and elevated AFP was found in 4 patients. Initial diagnosis based on elevated marker was established in 1 patient. In 7 cases, biopsy or partial resection was performed upfront. Initial complete resection was done in 3 patients. Preoperative chemotherapy according to TGM-95 was used in 8 patients. Only 3 patients received postoperative chemotherapy using the same cycles. Complete remission was achieved in 9 patients. Two patients with malignant teratoma and embryonal carcinoma died of progression. Mediastinal primary GCT are very rare. In children and adolescents with mGCT, the prognosis of most patients is favourable when complete resection is attained. Younger children appear to have a better prognosis. Patients with chemoresistant, unresectable tumours are at risk of poor outcome.

GCT-P13 Paediatric extracranial germ cell tumours in a tertiary hospital in Nigeria: Management challenges

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Background: Germ cell tumors are those tumors that arise from primordial pluri-potent cells destined to become either egg or sperm (gonadal). The extra-gonadal germ cell tumors develop along the pathway of germ cell migration to the gonads namely head, neck, mediastinum, retroperitoneum and sacrococcygeal region. They may be benign or malignant and comprise three main histological types: mature teratomas, immature teratomas and malignant germ cell tumour subtypes. Generally, cancer survival remains low in resource poor countries like ours due to several interrelated reasons like late presentation, diagnostic challenges, fake drugs, high cost of treatment, lack of trained manpower etc.
Methods: 4 representative cases are described which highlight the challenges.
Results: All 4 described cases presented late, in advanced stage and had incomplete treatment. These were attributable to ignorance, poverty, poor response to chemotherapy and lack of some treatment facilities (radiotherapy in our institution) and high cost of treatment. We therefore, recommend awareness creation to general public, encourage free treatment for childhood malignancies and collaborations/twinning with more developed centres.