RARE-58. CONGENITAL METASTATIC CHORDOMA OF THE CLIVUS

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Chordomas are rare midline axial skeletal neoplasms that typically present in adults. They are infrequent in childhood with typical localization in the spheno-occipital skull base. They are derived from remnants of the embryonic notochord. We present the case of 4 months old girl, who was born with „blueberry muffin" syndrome and was first negatively diagnosed for neuroblastoma and leukemia (two negative skin biopsies were performed) was admitted with axial laxity. In imaging testes there was a tumor of the scull base, metastases in the lungs and kidneys (that were not seen at previous assessments) and a small lesion in the heart. The third biopsy of skin lesion was performed and pathological examination revealed a neoplasm composed of cords, clusters, and chains of multivacuolated cells embedded within a myxoid matrix and separated by fibrous septa. No atypical and dedifferentiated features were present. Mitotic activity was not observed. Neoplastic cells showed the typical cytoplasmic immunostaining for EMA, S100 and cytokeratin AE1/AE3, strong nuclear brachyury expression, and retention of nuclear INI-1 expression. The diagnosis of chordoma was established. Neoplastic tissue and blood samples were obtained for molecular analysis using next generation sequencing, including germline mutations assessment (are ongoing). Chemotherapy as for soft tissue sarcomas was undertaken. Currently a patient is on treatment with improvement of neurological status.

RARE-59. CARDIAC REMODELING IN PATIENTS WITH CHILDHOOD-ONSET CRANIOPHARYNGIOMA – RESULTS OF HIT-ENDO AND KRANIOPHARYNGEOM 2000/2007

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BACKGROUND: Hypothalamic obesity caused by childhood–onset craniopharyngioma results in long–term cardiovascular morbidity. Knowledge about clinical markers and risk factors is rare. PATIENTS AND METHODS: A cross–sectional study on transthoracic echocardiographic parameters was performed to determine the associations with clinical and anthropometric parameters in 36 patients with childhood-onset adamantinomatous craniopharyngioma. RESULTS: Body mass index (BMI) correlated with the thickness of interventricular septum in diastole (IVSd) ($r=0.604$, $p<0.001$) and left ventricular diastolic posterior wall in diastole (LVPWd) ($r=0.460$, $p=0.011$). Due to wide range of disease duration, 17 pediatric and 19 adult patients were analyzed separately. In the adult subgroup (age at study ≥18 years), BMI correlated with IVSd ($r=0.707$, $p=0.003$), LVPWd ($r=0.592$, $p=0.020$) and left ventricular internal diameter in diastole (LVIDd) ($r=0.571$, $p=0.026$). In the pediatric subgroup (age at study <18 years), no correlation between cardiac parameters and BMI was observed. Only LVIDd correlated with disease duration ($r=0.645$, $p<0.001$). All cardiac functions were within the normal range, indicating no association with severe functional impairments. CONCLUSIONS: Cardiac remodeling in patients with childhood-onset craniopharyngioma correlates with the degree of hypothalamic obesity and disease duration. However, echocardiography has limited sensitivity in craniopharyngioma patients with obesity, so cardiac magnetic resonance imaging (MRI) should be considered as an alternative diagnostic approach for patients with craniopharyngioma and hypothalamic obesity.

RARE-60. PREGNANCIES AFTER CHILDHOOD CRANIOPHARYNGIOMA – RESULTS OF KRANIOPHARYNGEOM 2000/2007

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BACKGROUND: Data on female fertility, pregnancy, and outcome of offspring after childhood-onset craniopharyngioma (CP) are rare. STUDY DESIGN: Observational study on pregnancy rate and offspring outcome in female CP patients recruited in KRANIOPHARYNGEOM 2000/2007. RESULTS: A total of 451 CP patients (223 female) have been recruited, and 269 (133 female) were postpubertal at study. Six of 133 female CP patients (4.5%) with median age of 14.9 years at CP diagnosis had 9 pregnancies, giving birth to 10 newborns. Three patients achieved complete surgical resections. No patient underwent postoperative irradiation. Five natural pregnancies occurred in 3 CP patients without pituitary deficiencies. Four pregnancies in 3 CP patients with hypopituitarism were achieved under assisted reproductive techniques (ART) (median 4.5 cycles, range: 3–6 cycles). Median maternal age at pregnancy was 30 years (range: 22–41 years). Six babies (60%) were delivered by caesarean section. Median gestational age at delivery was 38 weeks (range: 34–43 weeks); median birth weight was 2,920 g (range: 2,270– 3,520 g), the rate of preterm delivery was 33%. Enlargements of CP cysts occurred in 2 women during pregnancy. Other complications during pregnancy, delivery, and postnatal period were not observed. CONCLUSIONS: Pregnancies after CP are rare and were only achieved after ART in patients with hypopituitarism. Close monitoring by an experienced reproductive physician is necessary. Due to a potentially increased risk for cystic enlargement, clinical, ophthalmological, and MRI monitoring are recommended in patients at risk. Perinatal complications, birth defects, and morbidity of mothers and offspring were not observed.

RARE-61. BODY COMPOSITION AND NUCHAL SKINFOLD THICKNESS IN PEDIATRIC BRAIN TUMOR PATIENTS

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BACKGROUND: Obesity, cardiovascular disease (CVD), and relapse/progression have major impact on prognosis in pediatric brain tumor (BT) patients. Cranial MRI is part of routine follow-up. METHODS: In a cross-sectional study, we analyzed nuchal skinfold thickness (NST) on MRI performed for BT follow-up monitoring as a novel parameter for body composition (BC) and CVD in 177 BT patients (40 WHO grade 1–2 BT; 31 grade 3–4 BT; 106 craniopharyngioma (CP)), and 53 healthy controls (HC). Associations of NST with body mass index (BMI), waist-to-height ratio (WHtR), caliper-measured skinfold thickness (cSFT), and blood pressure (BP) were analysed in BT and HC. RESULTS: CP patients showed higher BMI, WHtR, NST and cSFT when compared with BT and HC, whereas these differences were not detectable between BT and HC. However, WHO grade 1–2 BT patients were observed with higher BMI, waist circumference and triceps cSFT when compared to WHO grade 3–4 BT patients. NST showed high correlations with BMI, WHtR, and cSFT. NST, BMI and WHtR had predictive value for CVD in terms of increased BP, and in multivariate analysis, only BMI was selected for the final model resulting in an odds ratio of 1.25 (1.14–1.379). In CP patients with hypothalamic involvement/lesion or gross-total resection, rate and degree of obesity were increased. CONCLUSIONS: As monitoring of MRI and BC play an important role in follow-up after BT, NST could serve as a novel useful parameter for assessment of BC and CVD risk in BT patients.

RARE-62. VISUAL FUNCTION IN CHILDREN WITH CRANIOPHARYNGIOMA AT DIAGNOSIS: A SYSTEMATIC REVIEW

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Childhood craniopharyngioma is a rare and slow growing brain tumour, often located in the sellar and suprasellar region. It commonly manifests with visual impairment, increased intracranial pressure and hypothalamic and/or pituitary deficiencies. Visual impairment in childhood adversely affects a child's daily functioning and quality of life. We systematically reviewed the literature to provide an extensive overview of the visual function in children with craniopharyngioma at diagnosis in order to estimate the diversity, magnitude and relevance of the problem of visual impairment. Of the 543 potentially relevant articles, 84 studies met our inclusion criteria. Visual impairment at diagnosis was reported in 1041 of 2071 children (50.3%), decreased visual acuity was reported in 546 of 1321 children (41.3%) and visual field defects were reported in 426 of 1111 children (38.3%). Other ophthalmological findings described were fundoscopic (32.5%) and orthoptic abnormalities (12.5%). Variations in ophthalmological testing methods and ophthalmological definitions precluded a meta-analysis. The results of this review confirm the importance of ophthalmological examination in children with craniopharyngioma at diagnosis in order to detect visual impairment and provide adequate support. Future studies should