AT-16. CASE REPORT OF METASTATIC ATYPICAL TERATOID RHABDOID TUMOR (ATRT) SECONDARY TO AN INHERITED GERMLINE MUTATION IN THE SMARCA4 GENE: LITERATURE REVIEW AND RECOMMENDATIONS FOR TUMOR SURVEILLANCE OF ASYMPTOMATIC CARRIERS

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Biallelic inactivation of SMARCA4 has rarely been identified in rhabdoid tumor (RT) and atypical teratoid rhabdoid tumor (ATRT). SMARCA4-mutated ATRT is reported to have a higher frequency of inherited germline mutations and a worse prognosis compared to SMARCB1-mutated ATRT. The proband presented at 3.5 months of age with failure to thrive, left cranial III nerve weakness, and seizures. MRI of his brain and spine demonstrated a left thalamic mass with leptomeningeal metastases. A stereotactic biopsy of the thalamic mass demonstrated an embryonal tumor, with features of ATRT, but INI1 staining was positive for SMARCB1 protein expression. Additional immunohistochemical staining demonstrated loss of BRG1, consistent with biallelic inactivation of the SMARCA4 gene and diagnostic of SMARCA4-mutated ATRT. Genetic testing of his blood identified a single base pair deletion in the SMARCA4 gene (SMARCA4 c.2922del) resulting in a frame shift and premature stop codon, consistent with Rhabdoid Tumor Predisposition Syndrome-2 (RTPS2) (OMIM#613325). The family chose no treatment. The proband died of ATRT within 4 weeks of his diagnosis. The mother and grandmother, 22 and 49 years of age respectively at the time of genetic testing, were found to be asymptomatic carriers of SMARCA4 c.2922del. The mother subsequently became pregnant and genetic testing on the fetus identified the germline SMARCA4 c.2922del. The baby sibling of the proband is now 4 months of age and thus far has no evidence of ATRT or RT based on surveillance imaging. A review of the literature and recommendations for tumor surveillance in RTPS2 are presented.