DSS 2015 - 8

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Clinical History

- 26 year-old man with a 10 year history of right leg weakness:
- Three week history of urinary retention
Non-Contrast Magnetic Resonance Imaging
Diagnosis?
Differential Diagnosis

- Anaplastic ganglioglioma (WHO grade III)
- Atypical teratoid/rhabdoid tumor arising in ganglioglioma (WHO grade IV)
- Epithelioid glioblastoma with ganglioglioma-like foci (WHO grade IV)
• The tumor was negative for BRAF V600E and IDH1(R132H) immunostains and INI1 was retained in the tumor nuclei.
• There was no *EGFR* amplification or *PTEN* deletion.
Diagnosis

• Spinal glioblastoma with epithelioid features, ganglioglioma-like foci and $H3K27M$ mutation
Discussion

• Epithelioid glioblastoma
  – Primary glioblastoma
Discussion

• *BRAF V600E* mutation in ~50% of cases
• Negative for *IDH1 (R132H)* mutation (one exception reported)
• Negative for *EGFR* amplification
Peculiar Findings in Our Case

• Symptoms of long duration
• Ganglioglioma – like foci
• Loss of ATRX in the glioma-like areas but retained ATRX in the ganglioglioma-like areas
• \textit{H3 K27M} mutation
**K27M Mutation**


Common in thalamic high grade gliomas in young adults (Aihara K. et al. Neuro-Oncol. 2014;16(1),140-146)

Follow-Up

• The patient underwent radiotherapy and treatment with temozolomide
• Vertebral fracture due to osteonecrosis (radiotherapy-related)
• Currently he has stable disease and is back to work (six months follow-up)
Thank you
References