Bilateral Vestibular Schwannomas in a Patient with Ring Chromosome 22

Poster 2390

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Disclosure

• None
Introduction

• Ring chromosome 22 can lead to the development of intracranial tumors including meningioma, neurofibroma, schwannoma, mimicking NF-2

• Rare genetic disorder, truncation both ends of chromosome 22 with fusion of chromosome arms into a ring

• Lose tumor suppressor gene NF2
Methods

• We reviewed complete medical records, neuro-imaging studies, and genetic evaluation of young woman with ring chromosome 22 and bilateral vestibular schwannomas

• We reviewed available literature on this entity
Results

- 18 year-old girl with history of global developmental delay and cognitive impairment presents with sudden hearing loss
- MRI reveals bilateral vestibular schwannomas
- Audiographic monitoring shows progressive hearing loss
- Patient treated with Cyberknife SRS to larger tumor in ear with hearing loss, smaller tumor being monitored with audiometry and MRI
- Genetic evaluation and counseling for patient and family
Discussion

• Very rare genetic condition
• May predispose patients to bilateral vestibular schwannoma giving presentation identical to NF-2
• Genetic screening and counseling needed
• Recommend serial audiograms and MR imaging
• Goal to preserve hearing whenever possible
Summary

• Ring chromosome 22 is a rare genetic condition which can produce an “NF-2 phenotypic presentation” in young patients.
• Patients with ring chromosome 22 are at high risk for bilateral vestibular schwannoma.
• Hearing preservation should be goal whenever possible.

• Thank you!