



Novel Intracranial Imaging Findings in A Patient with Gorlin Goltz Syndrome

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Abstract

Gorlin Goltz syndrome, also known as Nevoid Basal cell Carcinoma Syndrome, is a rare entity with clinical neurologic manifestations of seizures. Imaging findings include falcine calcification, although there are recent reports of intracranial neoplasms, such as Meningioma.

Primitive neuro ectodermal tumors are noted in the minor criteria for establishing the diagnosis. We report the case of a twelve year female with imaging findings stable cerebral atrophy, cerebral white matter and brain stem signal abnormality suggestive of leukomalacia, and prominent virchow robin (perivascular) spaces. The case demonstrates novel central nervous system pathology in this rare disease.

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Introduction

Gorlin Goltz syndrome, or nevoid basal cell carcinoma syndrome, is a rare autosomal dominant disease with high penetrance caused by a genetic abnormality on the long arm of chromosome 9q22.3 in the area of the PTCH gene [1,2]. It may be sporadic in twenty percent of patients [3]. Clinical manifestations primarily affect the skin and the skeletal system however central nervous systems features have been described. Case reports have recently documented an association with meningioma and the literature places medulloblastoma (primitive neuroectodermal tumor) in the minor criteria [4,5].

Case description

We report a 12 year female who presents for initial MRI of the brain for seizure workup. Patient is a left handed female with nystagmus and partial symptomatic epilepsy with complex partial intractable seizures without status epilepticus. Electroencephalogram (EEG) classification is abnormal with a PDR of 9Hz. EEG demonstrates intermittent focal arrhythmic theta and delta slowing over the left posterior quadrant. Frequent sharp waves seen over the left occipital region (O1).

Her seizure history was initially documented at age 3; seizures were infrequent and untreated. The patient experience

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her first generalized tonic clonic seizure at age seven requiring hospitalization. Her neurological exam was significant for headaches and learning disability.

The patient was status post normal spontaneous delivery at 39 weeks gestational age to a 25 year female, gravida one. The pregnancy was uneventful.

The patient was discharged to home from the obstetric service but the family noted at 2 weeks that the patient was growing eyelashes, eyebrows and fingernails on her right side only. In addition, her left eye looked cloudy. She was diagnosed by the Ophthalmology service with glaucoma at age 3 months and underwent multiple surgeries. The patient is currently blind in the left eye. In addition, she has had cataract surgery on her left eye.

Dermatologic evaluation demonstrated multiple pigmented lesions primarily on legs and buttocks.

Genetics service ultimately diagnosed her with Gorlin Goltz syndrome at age 12. There was no documented family history of the disease.

The patient's dental history was significant for multiple cranial facial surgeries. The patient did have deciduous teeth, which when lost did not result in eruption of permanent dentition. The patient is to undergo dental implant surgery.

Magnetic Resonance Imaging (MRI) revealed cerebral atrophy but also extensive signal abnormality in the cerebral white matter and brainstem on T2 and FLAIR imaging consistent with leukomalacia. In addition, prominent perivascular spaces (Virchow-Robin) were noted within and adjacent to the leukomalacia. Similar appearing, but less pronounced, leukomalacia is noted within the brain stem (Figure 1).

Follow up imaging is performed three, ten and twelve years following initial MRI and revealed static findings of leukomalacia and prominent perivascular spaces (Figure 1).

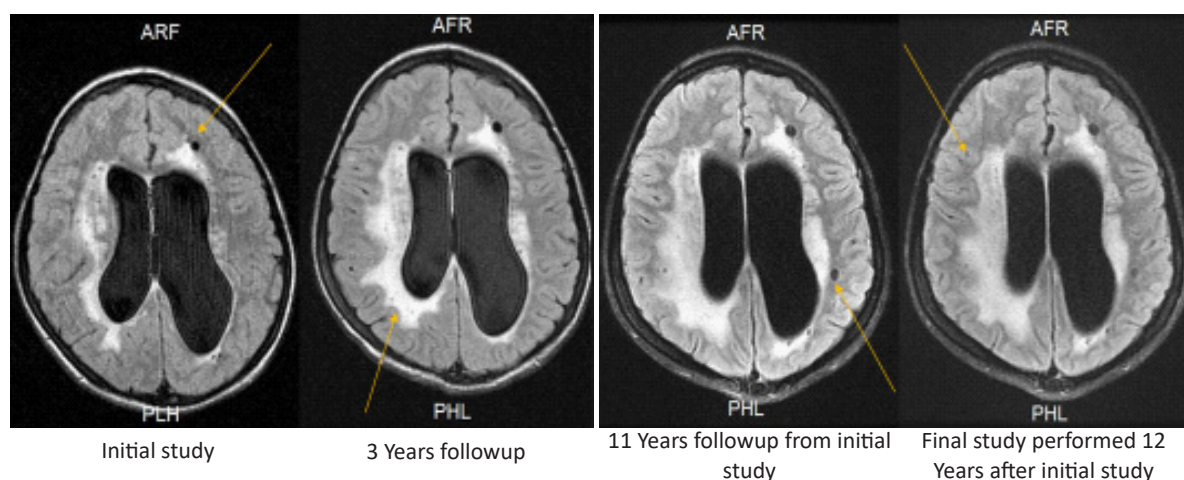


Figure 1: Serial axial T2 FLAIR images at the level of the corona radiata demonstrate stable intracranial stable cerebral leukomalacia and prominent virchow robin spaces (yellow arrows).

Discussion

Gorlin Goltz syndrome is a multisystem anomaly which primarily involves the cutaneous and skeletal systems [6,7]. It classically presents with odontogenic keratocysts, palmar and plantar pits with its predominant oncologic presentation basal cell carcinoma. Additional major criteria for the diagnosis of the syndrome include basal cell nevi, ectopic calcification, such as falcine calcification, and a family history of nevoid basal cell carcinoma.

Minor criteria include congenital skeletal anomalies, fibromas, primitive neuroectodermal tumor, and ocular anomalies, such as myopia, strabismus, cataract formation and glaucoma [4,8].

When involved, the central nervous system clinical manifestations include developmental delay, seizure disorder and hydrocephalus [9].

Intracranial imaging findings are rare, with falcine calcification the usual manifestation [1,9]. There is an association with intracranial neoplasia, with recent reports of meningioma resection [4]. Primitive neuroectodermal tumor is present in twenty percent of patients and is included in the syndrome's minor criteria [5]. There are two case reports of chiari malformations with hydrocephalus [10,11].

Our patient has novel intracranial findings including cerebral white matter leukomalacia with associated enlarged perivascular spaces. In addition, serial imaging over a twelve year period demonstrates static findings.

Conclusion

It is conceivable that continued imaging of the brain in this rare syndrome may yield additional imaging findings to further elucidate this syndrome which heretofore has primarily involved the skin and skeletal system.

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