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CASE REPORT

# A child with a congenital epidermal nevus, epilepsy and developmental delays: Linear nevus sebaceous syndrome

Carl E. Stafstrom[1] and Delora L. Mount[2]

***Abstract:***
This report describes a child who presented at birth with a large congenital scalp nevus that was successfully serially excised in three stages during the second year of life. As the child's development progressed, delayed milestones became apparent and seizures developed. The child was diagnosed with linear nevus sebaceous syndrome (LNSS). LNSS is a subtype of epidermal nevi associated with seizures, psychomotor retardation and neurocognitive delays. It is essential for pediatricians to be aware of the existence of a spectrum of epidermal nevus syndromes and make timely treatment referrals.

***Keywords:*** *linear nevus sebaceous syndrome, epidermal nevus, epilepsy, developmental delays, nevus of Jadassohn*
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## Introduction

Many infants and young children present for evaluation and treatment of congenital epidermal nevi. The overwhelming majority of these lesions are benign at the time of presentation, but because of potential for malignant degeneration later in life, removal is often recommended. In the majority of cases the excision proceeds uneventfully. However, some epidermal nevi are associated with specific syndromes and concomitant abnormalities in other areas such as the central nervous, skeletal, and ocular systems. Linear nevus sebaceous syndrome (LNSS) is a subtype of epidermal nevi associated with seizures, psychomotor retardation and neurocognitive delays [1-3]. LNSS is regarded as a sporadic disorder affecting approximately 1 in 1000-10,000 live births. It is presumably

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caused by genetic mosaicism with a lethal autosomal dominant gene [4].

**Case Report**

The Caucasian child was born at 39 weeks gestation following an uneventful pregnancy, labor, and delivery. Birth weight was 8 pounds, 1 ounce and there were no perinatal problems.

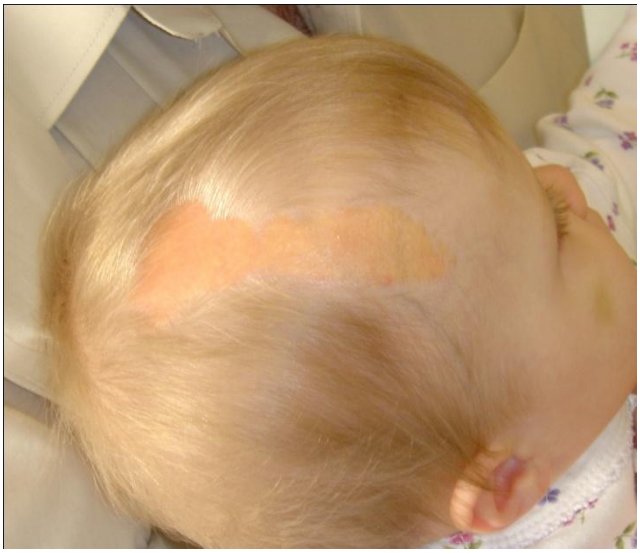

**Figure 1. Preoperative photograph of child with epidermal nevus.**

A large, a non-hair-bearing, raised, salmon-colored nevus was noticed at birth (Figure. 1). The lesion was located on the right fronto-parietal scalp and measured 70 x 28 mm.

Upon initial referral to the Pediatric Plastic Surgery service at age 10 months, the child had a normal head shape and no reported neurological concerns.Serial excision was performed in 3 stages, from 12 to 22 months of age, under general anesthesia. No intraoperative or perioperative complications were encountered and the site healed appropriately. Surgical pathology showed a typical nevus of Jadassohn without evidence of atypia or dysplasia (Figure 2).

Developmentally, the child began walking at 11-1/2 months. However, by about 14 months of age, the parents became concerned about developmental delays, particularly with regard to expressive and receptive language. Also, at 14 months of age, the child developed intermittent jerks of the head and body in clusters, especially during drowsiness. There was no family history of epilepsy, developmental delays, or congenital nevi.

Initial evaluation by a pediatric neurologist at age 15 months revealed no dysmorphic features other than macrocephaly (head circumference just above 98$^{th}$ percentile for age).

The remainder of the general examination was normal. On neurological examination, there were no deficits of cranial nerves 2-12. Cranial nerve 1 was not tested. The child had normal elemental sensory and motor function and an age-appropriate, stable gait. Fine motor coordination was adequate for age with well-developed pincer grasps bilaterally. Tendon reflexes were normoactive and plantar responses were flexor. However, delays were apparent in speech, language, and cognitive function. At the initial evaluation, the child was nonverbal except

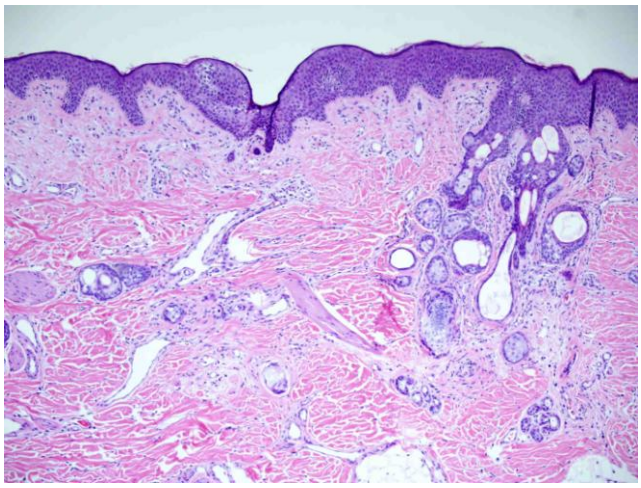

**Figure 2. Surgical pathology slide of resected lesion, consistent with nevus of Jadassohn. Skin shows absence of terminal hairs and malformed pilosebaceous complex with dilated empty hair canals. (Hematoxylin and eosin x100)**

for squealing vocalizations. The child had a poor attention span, had little interest in toys, did not point to body parts, and did not give objects to the examiner or parent on request.

Investigations included an EEG, which showed frequent epileptiform bursts of polyspike-wave discharges emanating from the right posterior quadrant (temporal and occipital lobes), the same hemisphere as the cutaneous nevus. Magnetic resonance imaging (MRI) scan of brain was normal with no dysgenesis, malformation, delay in myelination, or hydrocephalus. The child was started on an anticonvulsant (valproic acid), with marked decrease in myoclonic seizures. Subsequently, seizures of several types (partial, myoclonic, and generalized tonic-clonic) developed and became refractory to valproate. Recently, seizures have been well-controlled on dual therapy with oxcarbazepine and lamotrigine with no witnessed seizures for the past 18 months.

The child's subsequent development remained significantly delayed. At the age of 29 months, the child has about 10 single words of speech, many unclear and used inconsistently. Autism spectrum disorder was diagnosed at 2-1/2 years of age. Attention span is poor and the child is easily distractible with tactile defensiveness. The child receives early intervention services including speech therapy. There has been no developmental regression or plateau though progress has been slow. The child's head is growing along a stable trajectory above the 98$^{th}$ percentile.

**Discussion**

Epidermal nevi of the Jadassohn type are congenital skin lesions composed of hamartomatous tissues derived from epidermal, follicular, sebaceous and apocrine gland tissue. Nevi are usually excised due to cosmetic considerations (approximately 50% occur on the scalp as alopecic lesions) [5] as well as their proclivity to develop secondary malignant neoplasms later in life [6,7]. Early excisional treatment of nevus sebaceous lesions is controversial, as it is rare for malignant degeneration to occur in childhood. Lifetime risk of conversion of a nevus sebaceous lesion to basal cell carcinoma or squamous cell carcinoma ranges from 5-22% [8].

Linear nevus sebaceous syndrome (LNSS) has a number of associations in addition to cutaneous nevi. Commonly involved systems include skeletal (hypophosphatemic rickets), ocular (colobomata), and brain (seizures, developmental delays, and mental retardation) [9]. The cause of LNSS is unknown and there do not appear to be any unique genetic abnormalities or exogenous etiologies such as infection, trauma, or radiation. The reported incidence is 1 in 1000-10,000 births, without clear sexual predilection [7]. Many nevi have genetic mosaicism [10]. It has been hypothesized that LNSS, which involves structures of ectodermal and mesodermal origin, may be caused by anomalous development of the neuroectoderm prior to the fourth week of gestation, resulting in abnormalities of the brain, eyes, and skeleton [11].

Epilepsy, involving both generalized and partial seizures, is common in LNSS, afflicting 30-75% of affected children [9, 12]. Seizures can become refractory to medical treatment. Other neurologic manifestations include psychomotor retardation and neurocognitive delays [13-16]. Some affected children have neuronal migration disorders or other brain structural anomalies such as hemimegalencephaly with associated facial hemihypertrophy [17]. Several patients with LNSS and hemimegalencephaly have undergone hemispherectomy for seizure control [18]. Macrocephaly is common, sometimes related to underlying brain structural anomalies or hydrocephalus. Prognosis of LNSS is variable, with cognition ranging from normal to profound impairment.

The surgeon and pediatrician should be attentive to developmental issues over the time period of

treatment of a nevus sebaceous of Jadassohn, in case a syndromic condition is present. Certainly, timely referral should be made if the question of seizure, other atypical movements, or developmental delay is reported by the family.

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