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Down syndrome and incidental pediatric basal ganglia calcifications at autopsy

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Abstract

Basal ganglia calcifications in the pediatric population are an unusual finding and in radiologic studies, have been noted in between 1-2% of patients in that age group. The causes for calcification are myriad but most commonly include tumors, congenital infections and tuberous sclerosis. The current report describes a case of a 17-year-old female with Down syndrome (trisomy 21) who presented with agitation, dry cough, decreased appetite, and fever. She experienced a 3 minute apneic episode followed by cardiopulmonary resuscitation. She was noted on imaging studies to have bilateral opacities in the lungs with consolidation. She was started on antibiotics. She died and at autopsy, was diagnosed with panlobar bronchopneumonia. Angiocentric calcifications were incidentally noted in the basal ganglia. This report will present a brief review of the literature on pediatric basal ganglia calcifications and their association with Down syndrome.

Keywords: Calcifications; Basal ganglia calcifications; Pediatric; Down syndrome; Trisomy 21

Introduction

There is a whole host of conditions which may be associated with the development of calcifications in the basal ganglia region [1,2]. These conditions run the gamut from aging to metabolic disease (parathyroid disorders), tumors, infections (congenital infections, tuberculosis, AIDS, neurocysticercosis, toxoplasmosis), toxic conditions (carbon monoxide poisoning, lead poisoning), radiation and chemotherapy, inherited disorders (mitochondrial disease, Cockayne syndrome, Pantothenate kinase associated neurodegeneration, tuberous sclerosis), vascular developmental venous anomaly, Fahr disease, and birth hypoxia. Anecdotal cases of basal ganglia calcifications have also been documented in association with Down syndrome at autopsy [3]. We report a case of a 17-year-old female with Down syndrome who died of panlobar bronchopneumonia and who had incidental microcalcifications noted in the basal ganglia at the time of autopsy.

Case report

The patient was a 17-year-old female with a history of Trisomy 21 (Down syndrome) and associated developmental delay. She was born the product of a full term pregnancy with no delivery complications. At age 2.5 months, she underwent ligation of a patent ductus arteriosus due to failure to thrive and congestive heart failure. At age 3.5 months, she underwent an anterior cricoid split due to subglottic stenosis and associated respiratory distress. Most recently, she presented to the emer-

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gency room with agitation. Six days prior, her mother noticed that she had a dry cough, decreased appetite and lethargy. She received Lorazepam and Diphenhydramine. Subsequently, she experienced a 3 minute apneic episode and cardiopulmonary resuscitation was started. She was empirically started on Vancomycin and Piperacillin-Tazobactam following a chest X-ray, which showed multifocal bilateral opacities in the lungs and a Computed Tomography study (CT) which showed bilateral lower lobe consolidation. Worsening hypoxia and hypotension prompted a CT study of the head which showed changes suggestive of possible anoxic damage. She subsequently expired and an autopsy was performed.

At autopsy, she was noted to have panlobar bronchopneumonia with intraalveolar hemorrhages (Figure 1). Serous pleural effusions were also seen bilaterally. Examination of the brain showed focal dystrophic calcifications in the basal ganglia (Figure 2); most of the calcifications appeared to be associated with small blood vessel walls. Dystrophic calcifications were not observed elsewhere in the central nervous system. Foreshortening of the frontal lobes was also noted; the brain weight was 989 grams.

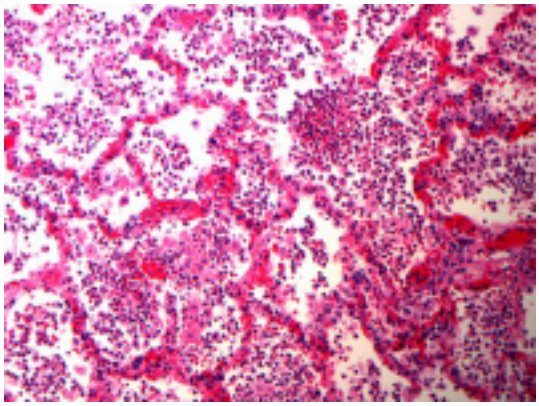

Figure 1: All lobes of lung bilaterally showed intraalveolar acute inflammation consistent with acute bronchopneumonia (hematoxylin and eosin, original magnification 200X).

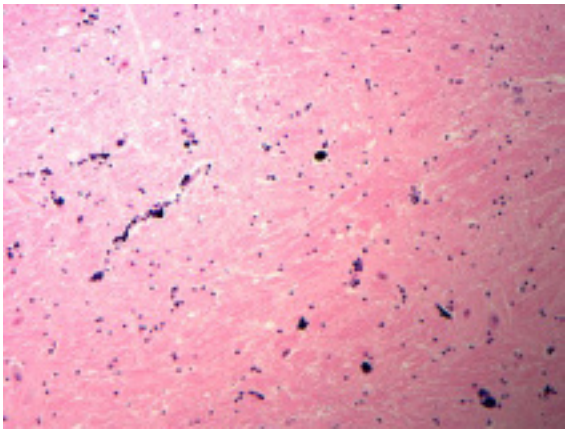

Figure 2: Microcalcifications were noted focally in the basal ganglia (hematoxylin and eosin, original magnification 200X).

Discussion

Basal ganglia calcifications are an infrequent finding in the pediatric population. Legido and colleagues in reviewing 6428 head Computed Tomography (CT) scans done on 4283 children, found evidence of basal ganglia calcification in 48 children (1.1%) [4]. The most common etiologies for the calcification in their series were tumor, tuberous sclerosis and history of a congenital infection. A number of patients reported in this series had developmental delay but the presence of calcifications did not appear to be directly related to a specific form of neurologic dysfunction. The authors conjectured that the presence of calcifications was a marker for brain damage. In the current case, the patient had a history of symptomatic patent ductus arteriosus and subglottic stenosis, both necessitating surgery. Both of these conditions may have resulted in anoxic damage to the brain and may have contributed to the development of the observed calcifications.

Stakahima and Becker reviewed an autopsy series of 33 Downs syndrome patients who were older than 1 year of age (age range 1-60 years) and who had good representative sections of basal ganglia available for examination [3]. Thirteen patients (39.4%) had definitive evidence of micro calcifications. Of those, 4 were pediatric patients. Three of the four pediatric cases had what was described as “mild” calcifications defined as single globules in all microscopic fields. The fourth pediatric case had severe calcification with massive deposits. Of the nine adult patients in the series, seven had moderate to severe calcification. An additional nine pediatric cases had what was described as “borderline” calcification with single globules in some microscopic fields. Similar to the current case, most of the calcifications were observed in proximity to blood vessels. Additional clinical information regarding patient comorbidities, family history and clinical presentations or course were not provided in this study.

The pathogenesis of the calcifications in the setting of Down syndrome is not well understood. Wisniewski et al noted that the calcifications were more common in the youngest and oldest patients [5]. They postulated that calcifications in the younger patients may be related to ischemia in the brain and that this might be secondary to other comorbidities (such as heart disease) in this population of patients. Okano et al alternatively hypothesized that the early appearance of basal ganglia calcifications and brain atrophy are manifestations of accelerated aging in Down syndrome [6].

Identification of calcifications in childhood on imaging or at autopsy are unusual and as such, serve as a nonspecific marker for a host of potential underlying neurologic disorders, Downs syndrome included. In the current case, it is impossible to determine if the calcifications are intrinsically related to the fact the patient has Downs syndrome or whether they are secondary to possible hypoxic damage sustained early in her life.

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