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A Unique Case of Amelia and Congenital Scoliosis in a Vietnamese Female

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Abstract

Amelia and scoliosis are two types of skeletal deformities that can occur in vertebrates. Amelia is a rare birth defect in which bones of the extremities are completely absent. This condition has been linked to genetic inheritance as well as in utero exposure to thalidomide, an immunomodulatory drug. Scoliosis can be congenital or acquired. While known causes of scoliosis include cerebral palsy and muscular dystrophy, etiologies of the majority of cases are unknown. Complications of scoliosis can be grave when the spinal curvature is severe enough to affect other organ systems, necessitating treatment. We herein report an unfortunate case of amelia paired with congenital scoliosis in a patient with minimal access to healthcare. Interestingly, the patient's mother was potentially exposed to high environmental levels of dioxin, a component of the pesticide Agent Orange, but no other risk factors were discovered. The patient and her family are from a rural province of Vietnam, and do not have the ability to regularly seek medical caremaking treatment of her disease difficult. Given the detrimental effects on overall quality of life combined with the patient's inability to receive appropriate care, we report this case in an effort to not only increase awareness about a rare condition but also to shed light on barriers to treatment in impoverished nations.

Introduction

Amelia is an anomaly of the skeletal system that is particularly rare, with a reported prevalence of 1.41 per 100,000 births.[1] The combination of amelia with an additional congenital defect is increasingly uncommon, with a prevalence of .77 per 100,000.[2] Scoliosis is another anomaly of the skeletal system that can be congenital in nature. Both conditions represent birth defects whose etiologies remain largely unknown.[3,4] Malformations related to the skeletal system may adversely affect a patient's health and well-being. For example, when severe, scoliosis can lead tolife threatening deficits in both cardiovascular and respiratory function.[5]In addition, amelia of the upper extremities can create a significant reduction in the ability to perform activities of daily living such as bathing, feeding, and cooking, making everyday life difficult without assistance. Treatment of a severe scoliotic curvature requires surgical intervention, which may not be readily available for patients in rural areas with underdeveloped healthcare systems. Due to the patient's lack of access to healthcare and theoverall reduction in quality of life she has experienced from these conditions, we are led to report a case of amelia paired with congenital scoliosis in a Vietnamese female with no remarkable risk factors.

Case Report

In October of 2002, a Vietnamese female was born via vaginal delivery to a22-year-old mother. The child was a premature infant, being delivered early in the mother's third trimester secondary to painless vaginal bleeding. The etiology of the bleeding is unknown. Throughout delivery, the vital signs of the fetus remained stable, and there were no complications. After delivery, physical exam was remarkable for a newborn with complete absence of the upper limbs, paired with a severe scoliotic curvature, defined by a Cobb's angle greater than 40 degrees. Osteopathic structural examination was remarkable for a dextroscoliotic curve, side bent left, rotated right, involving the entire thoracic and lumbar regions. The diagnosis of idiopathic congenital scoliosis paired with bilateral amelia of the upper extremities was made. Due to the potential for other serious congenital anomalies, further imaging and a complete physical exam were performed.

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Ultrasound of the abdomen revealed no anomalies of the genitourinary and gastrointestinal systems, and echocardiography showed no evidence of congenital cardiac defects. Physical exam was only remarkable for the amelia and scoliosis. The external genitalia were normal and neurologic exam did not elicit any focal neurologic deficits. Vital signs remained stable following the workup and she was able to be discharged after routine postpartum care. Further investigation was sought regarding the course of the pregnancy, but no obvious causes for the malformations were discovered.

Upon further questioning, it was found that the fetus was not exposed to any known teratogenic agents during the pregnancy. The mother did not suffer from gestational diabetes nor hypertension. There was no history of infection during the pregnancy, and both the mother and the father were healthy, with unremarkable past medical histories. Furthermore, there was no family history of genetically inherited conditions. The child's parents and younger sister were full term with no anatomical malformations. Interestingly, the child was born in the Kon Tum province of Vietnam, which is known for having high environmental levels of dioxin, a component of the pesticide Agent Orange.

As the patient grew, the scoliotic curvature worsened and was accompanied by back pain. Given the increasing severity of symptoms, medical intervention became necessary. In 2015, at thirteen years of age, she underwent spinal surgery to improve her condition. The patient had bilateral growing rods implanted with pedunculated screws. The role of insertion of the growing rods was twofold, serving to straighten the abnormal spinal curvature and allow for further growth as the child aged.

Currently, the patient remains in rural Vietnam and continues to suffer from the severe scoliotic curvature, with a measured Cobb's angle of 54 degrees in the thoracic region, and 65 degrees in the lumbar region. Her family is applying for further treatment through Shriners Hospitals, a non-profit medical organization that treats children with orthopedic conditions regardless of their ability to pay. If approved, the patient will travel to the USA and undergo additional treatment for her medical condition in an attempt to improve her quality of life.

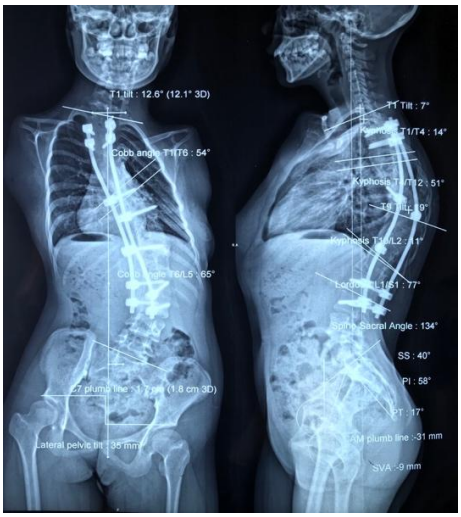

Figure 1: X-ray of the patient showing the scoliotic curvature. Included are Cobb's angles of the thoracic and lumbar spine.

Figure 2: Image of the patients back, showing the physical appearance of the scoliotic curve.

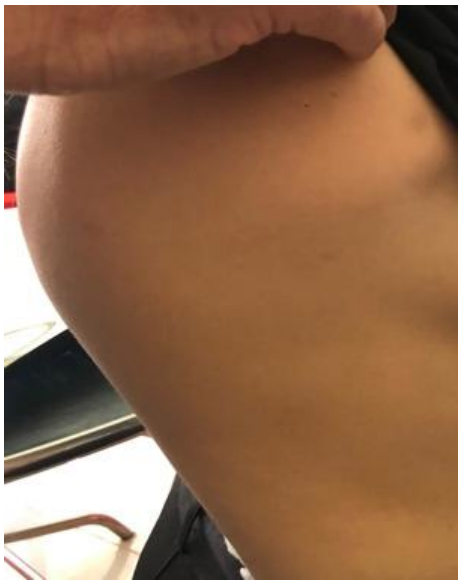

Figure 3: Image of the patient's thorax, showing the absence of her right upper extremity.

Figure 4: Image showing the bilateral amelia of the patient.

Discussion

Amelia is a rare birth defect that is characterized by the absence of one or more limbs.[1] This defect can be sporadic or secondary to toxin exposure.[4] Thalidomide, alcohol, and maternal diabetes have all been reported to cause this defect.[9,10,11] In rare cases, the birth of a child from consanguineous parents can lead to this deformity.[12]

There are three known mechanisms that can lead to a limb deficiency: malformation of the limb due to genetic mutations or an insult during formation of the blastomere, lack of blood flow to the developing limb, and limb amputation via the amniotic band.[1] Amelia can present alone or with other anomalies, including defects of the abdominal wall and/or kidneys.[4]Depending on the number of extremities affected, the condition can have a significant impact on lives of affected individuals by hindering their ability to perform activities of daily living.

Congenital scoliosis is a deformity of the spine that is defined by a longitudinal and rotational imbalance caused by abnormalities in the formation and segmentation of the spinal column during somitogenesis.[6,7] This condition has been linked to other disease processes including cerebral palsy and muscular dystrophy. No single etiologic agent has been found to cause this defect, but it has been postulated that carbon monoxide exposure during somite formation may lead to malformation.[3,7] In addition, maternal diabetes, fetal hypoxia, and various antiepileptic agents have been listed as potential causes.[7,8] Congenital scoliosis is also associated with anomalies of other organ systems including the nervous system (35% of cases), the cardiovascular system (25% of cases), and the urologic system (20% of cases).[6] Complications of congenital scoliosis may be life threatening depending on the severity of disease.[5] Based on the degree of curvature, possible treatment modalities are observation, bracing, or surgery.[3] Cases with a Cobb's angle greater than 40 degrees require surgical treatment.[6]In these cases, the severe curvature of the spinemay limit the expansion of the lungs and compress the chambers of the heart, thus

making surgical treatment a necessity.[5] The goal of each treatment is to lessen the abnormal curvature, improve patient mortality, and prevent disease progression.[3]

Dioxins are a group of highly toxic chemical compounds that have been considered persistent environmental contaminants due to their prolonged use throughout the 1900s.[13]The only remarkable information in the past medical history of our patient is potential in utero exposure to dioxins secondary to high environmental levels. There is a considerable amount of scientific research on dioxins and their potential to cause various birth defects. Exposure to dioxins during pregnancy has been shown to cause teratogenic effects in animal studies, however, human studies linking birth defects to dioxin exposure have been controversial.[14] There are no studies that directly link dioxin exposure to amelia or scoliosis, however, most studies related to its potential to cause birth defects have significant heterogeneity and small study populations.[14] More scientific research is needed in order to confirm the potential effects that dioxin has on human development.[14]

This case presents a child with congenital scoliosis combined with amelia of the upper extremities, born to a non-consanguineous couple in rural Vietnam. Organic causes of these conditions were ruled out. There was no remarkable toxin exposure during the pregnancy, other than being born in a region with a high environmental level of dioxins, which is controversial. Thalidomide, antiepileptics, alcohol exposure, as well as gestational diabetes were ruled out as potential causes. Genetic inheritance may also play a role in these birth defects, however, given the normal musculoskeletal anatomy of the child's immediate family combined with a negative family history of these defects, genetic inheritance is unlikely.[12] Commonly, these conditions may present with anomalies of other organ systems, including the nervous system, cardiovascular system, and urologic system.[6] This child presented with no defects relating to the systems listed above, which is a rare finding.

Severe scoliosis with a Cobb's angle greater than 40 requires surgical intervention due to potential cardio respiratory complications and secondary back pain. If the patient's cardiorespiratory functions are stable, the surgery is not considered emergent, and when surgery is deemed non-emergent, it can take many months to years before the patient is able to be treated in Vietnam. Additionally, the type of surgical treatment our patient needs is only available in the two major cities of Vietnam, Hanoi and Ho Chi Minh, both of which are over 12 hours away from her residence. The region of Kon Tum, where our patient resides, is a rural and impoverished mountain town. The access to healthcare is minimal and travel to these major cities takes many hours and can be expensive. Furthermore, the cost of the corrective surgery is also expensive and exceeds what the family could afford to pay, creating another barrier to treatment.

Infants born with scoliosis or amelia as an isolated finding generally have a good prognosis, but the combination of these two conditions increases the likelihood for complications after birth andcan have a significant impact on the quality of life of affected patients.[5]For example, our patient needs assistance with many of her activities of daily living such as bathing, cooking, and eating due to the absence of her upper extremities. In addition, she learned how to write with her feet, allowing her to advance in her academics. Fortunately our patient was able to have a partially corrective surgery in the past, however, as her need for additional surgery grows, access and cost remain active concerns for the family. Due to the potential complications of severe scoliosis and the reduction in the quality of life that amelia may cause, we present this case in order to emphasize how access to healthcare can make a tremendous impact on treatment various diseases.

To whom it may concern,

My name is Samuel Stahly and I am a 4th year medical student at the Arkansas College of Osteopathic Medicine. I am submitting this case report regarding a patient that I saw while on a mission trip in Viet Nam. The other authors of this paper are Bryce Sarcar and Dr. Stanley Grogg. I have provided their emails and contact information at the end of this letter. The goal of this paper is multifactorial. We wanted to share this case not only to provide further education on a rare medical condition, but also to shed light on barriers to the access of healthcare. This case is unique in that the social aspects of medicine had a direct impact on the care of the patient. We think this case would be a great fit for your journal due to the above mentioned reasons.

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