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BRUCK SYNDROME: A CASE REPORT

Buklaev D.S.[1]***, Kostik M.M.***[2]***, Agranovich O.E.***[1]***, Trofimova S.I.***[1]

[1] The Turner Institute for Children's Orthopedics, Saint-Petersburg, Russian Federation
[2] The Ministry of Health of Russia, St. Petersburg Saint-Petersburg State Pediatric Medical University

The article describes the clinical case of an infant with Bruck syndrome. The clinical and radiological analyses showed the presence of systemic osteoporosis with pathological fractures; contractures of the elbow, knee, and ankle joints; delay of physical and motor development; and signs of hypoplasia in some of the muscle groups. There was also a right-sided congenital muscular torticollis. X-ray analysis revealed a moderate antecurvation deformity of the lower legs and femurs, with cortical thinning. Laboratory data detected an abnormal beta-cross lap increase.Treatment of osteoporosis by inhibitors of osteoclastic resorption (pamidronate) had a positive effect, and the elimination of flexion contractures at the elbow using plaster bandages with the distraction device also resulted in a positive effect.

Keywords: Bruck syndrome, joint contractures, osteoporosis, children.

Bruck syndrome is an extremely rare congenital disorder with an incidence of less than $1/10^6$. This peculiar illness combines symptoms of osteogenesis imperfecta and congenital joint contractures, which has similar symptoms as arthrogryposis. Alfred Bruck first described the symptoms of congenital bone fragility and congenital contractures of the large joints in 1878 (1). Individual observations with similar manifestations have been reported in the literature (2-4).

During the last decade, genetic research has been conducted in an attempt to determine the mutations that may cause Bruck syndrome. Mutations were identified that result in defects of the primary collagen chain and its post-translational modifications (5-8).

The most commonly used main treatments include bisphosphonates to control osteoporosis and orthopedic therapy to correct positioning of limb segments and improvement in their function.

Case Report

We report a case of a patient with clinical and radiological symptoms of Bruck syndrome. The patient had a history of frequent bone fractures during the perinatal period, and later in infancy, limb deformities and joint contractures were observed. Bone fractures occurred even without adequate trauma. The child had a birth weight of 980 g at 30 weeks of gestation. The early neonatal period was difficult because of prematurity, fractures, and bronchopulmonary dysplasia. There is no family history of hereditary orthopedic pathology. The parents are healthy, and the marriage was not consanguineous.

To reduce osteoporosis and prevent fractures, since the age of 3 months, the patient received pamidronate, an osteoclastic resorption inhibitor, every 3 months at the Saint Petersburg State Pediatric Medical University clinic. The patient was examined at the Federal State Budgetary Institution "The GI Turner Scientific Research Institute for Children's Orthopedics" for the first time at the age of 10 months. The parents complained about upper and lower limb dysfunction due to elbow and knee joint contractures.

A physical examination demonstrated delayed physical and psychomotor development. In particular, the body weight was 7300 g and length was 68 cm, which corresponds to the 5th percentile on US Pediatric CDC Growth Charts. The body position was forced, with the child lying on his back, and the patient was able to hold his head upright. He could independently sit and roll over but could not stand because of low muscle tone and contractures. The gaze was steady, and he monitored his surroundings. He recognized his parents, and his emotional reaction to examination was active.

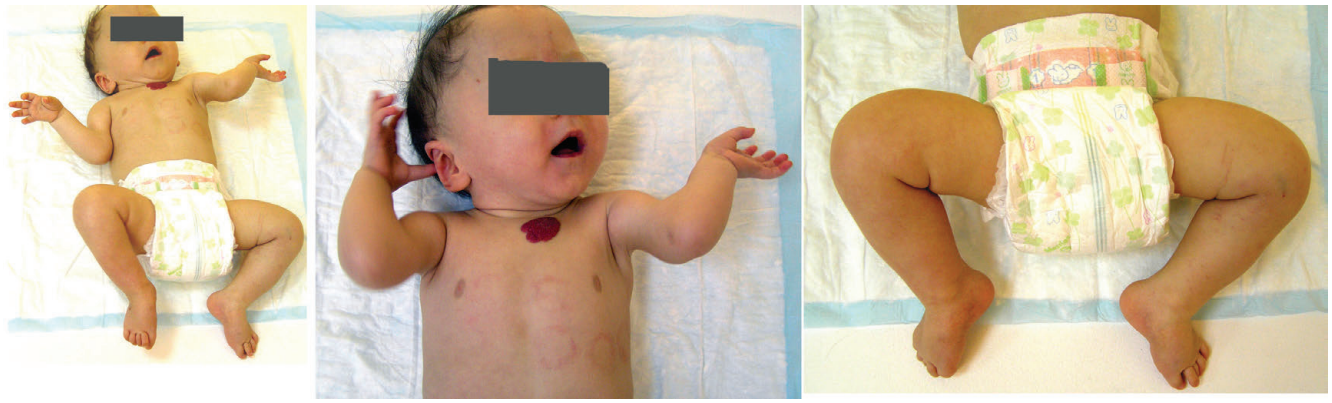

Fig. 1. Patient with Bruck syndrome

Cranial nerves were without focal symptoms, and the fontanelle was large (dimensions, 1.5 × 1.5 cm).

The head position was forced; it was turned to the left and tilted to the right. Accordingly, the rotation of the head to the left and tilt to the right were limited. The neck appeared shortened. Muscle palpation revealed a contracted and shortened left sternocleidomastoid muscle. The cranial shape was close to rhombic in the horizontal plane because of craniotabes and a prolonged forced position. Dentation was delayed. The sclerae were normal in color, which is characteristic of healthy babies, and did not reveal any expressed blue or gray color, which is characteristic in children with certain types of osteogenesis imperfecta.

The spine and chest had no visible deformities. There was insufficient strength and tone of the body muscles. The skin of the anterior surface of the chest had a 2.5 × 4 cm hemangioma with a tendency for regression. In general, contours of the shoulder girdle and upper limbs were intact. There was movement in the shoulder joints but with some restriction compared with normal movement.

The position of the forearms was abnormal because of flexion and pronation contractures in the elbow joints. The maximum active extension of the elbow was up to 80° for the right arm and up to 75° for the left arm. The only active movement the child could perform in the elbow joints was extension. Flexion was only passive and because of tissue elasticity. Palpation revealed a pronounced hypoplasia of the biceps on both shoulders, possibly even complete absence of functionally active muscle tissue. When the child was in a lying position, the forearms were in pronation, even excessive. Supination was only passively possible and only to the neutral position. Movement from the neutral position to supination, even incomplete, was impossible.

In general, the hands were properly developed. Symmetric intermittent ulnar deviation in the wrist, which was no more than 20°, was observed. The child could actively place the hand into the middle position; however, placement in the radial direction was only passively possible and to no more than 10°. Moreover, thenar muscle hypotrophy and mild adduction contracture of the first finger were observed but with virtually no effect on the function of the hands.

The lower limbs were in the middle position and had the same length and correct axis in the frontal plane. The range of motion in the hip joints was within the age norm. There were active leg movements. The knee joint extension was deficient (flexion contracture). Active extension was possible to an angle of 90° on both sides, with passive extension up to 100°. The feet were well developed. An equinus contracture of 110° was present on both the sides. Furthermore, feet contractures were rigid.

X-rays revealed no abnormalities and dysplasia of the skeleton. The position of the limb joints corresponded to the clinical picture. “Zebra lines” were observed in the metaphyses of the long bones and in the pelvis apophyses because of pamidronate administration. There was marked cortical thinning of the long bones; anteflexion deformation of the femur and tibia bones; and expansion of preparatory calcification zones, including in the vertebral apophyses. The cancellous structure of the vertebrae was blurred, and platyspondyly was present.

Laboratory tests were conducted. Among the detected abnormalities, only increase of beta-crosslaps was noted, which is apparently not a sign of the disease but rather the expected manifestation of an-

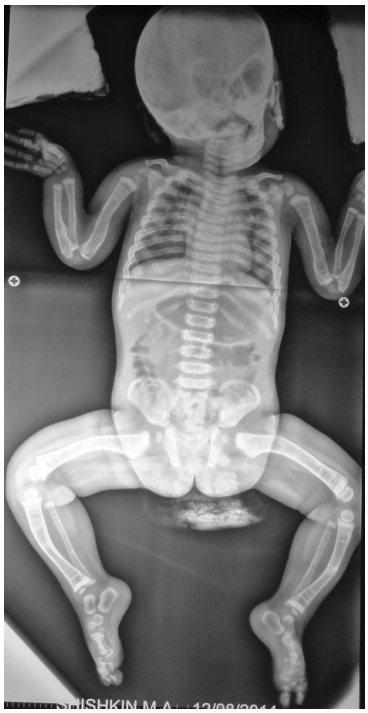

Fig. 2. X-ray of the patient with Bruck syndrome

tiosteoclastic therapy that causes the shift of osteosynthesis/osteolysis balance toward osteolysis. Thus, our patient had increased fragility of bones because of congenital osteoporosis and multiple congenital contractures. Therefore, we were inclined to consider this as a case of Bruck syndrome.

With respect to the treatment of the patient, osteoporosis therapy was the first priority, which would continue for another 3 to 4 years; this is similar to the treatment of osteogenesis imperfecta. For orthopedic treatment, gradual elimination of contractures of the elbow, knee, and ankle joints, followed by motor rehabilitation would be performed. At the time of publication, the first stage of distraction redressing of the elbow contracture in a cast had been performed, resulting in a decrease in flexion contractures to a 160°.

Discussion

Our case demonstrated clinical and radiological manifestations and the developmental course of a combination of two disorders, i.e., osteogenesis imperfecta and arthrogryposis. From the characteristic manifestations of osteogenesis imperfecta, we note an increased susceptibility to pathological fractures, even without adequate trauma and without clear symptoms; characteristic bone deformities of the lower limbs; platyspondyly; osteoporosis; and a good response to antiresorption therapy (decreased incidences of fracture). At the same time, such a significant delay in motor development in children with osteogenesis imperfecta, who regularly receive treatment with bisphosphonates, is not typical.

The type and nature of contractions in this case were typical for arthrogryposis. Contractures were rigid and muscle hypoplasia was present, which involved the same muscle groups as in cases of arthrogryposis. Contractures are myo-fibrous-arthritic in nature and are amenable to correction, although with great difficulty with the use of casts and a distraction device. In this manner, we managed to reduce flexion contractures of the elbow joints to an angle of 150°. An indirect confirmation of our assumptions regarding the nature of contractions was presented by Bank et al. (7), who noted that in Bruck syndrome, the structure of collagen in cartilage tissue is intact, i.e., the cause of contracture is not a primary defect of the articular cartilage, but changes in the muscles and connective tissue structures.

Conclusion

We report a clinical case of a rare condition, Bruck syndrome, which is a little studied disorder and requires further study. Treatment of patients should be of combined nature, with bisphosphonates drug therapy aimed at reducing osteolytic processes, orthopedic interventions to correct contractures of joints, and subsequent motor rehabilitation.

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СИНДРОМ БРУКА. ОПИСАНИЕ СЛУЧАЯ

© Буклаев Д.С.[1]***, Костик М.М.***[2]***, Агранович О.Е.***[1]***, Трофимова С.И.***[1]

[1] ФГБУ «НИДОИ им. Г. И. Турнера» Минздрава России, Санкт-Петербург
[2] ГБОУ ВПО «Санкт-Петербургский государственный педиатрический медицинский университет», Санкт-Петербург

В статье приводится описание клинического случая синдрома Брука у пациента грудного возраста. Клинико-рентгенологическая картина демонстрирует наличие системного остеопороза с патологическими переломами, контрактуры локтевых, коленных, голеностопных суставов, задержку физического и двигательного развития, признаки гипоплазии некоторых групп мышц. Также имеется правосторонняя врожденная мышечная кривошея. При рентгенографии выявлены умеренная антекурвационная деформация голеней и бедер, истончение кортикального слоя. Лабораторные данные показали отклонения от нормы только со стороны бета-кросслапс в сторону увеличения.

Проводится лечение по поводу остеопороза ингибиторами остеокластической резорбции (памидронатом) с положительным эффектом и сгибательных контрактур локтевых суставов гипсовыми повязками с дистракционным устройством также с положительным эффектом.

Ключевые слова: синдром Брука, контрактуры суставов, остеопороз, дети.

Information about the authors

Buklaev Dmitry Stepanovich — MD, PhD, chief of the department of arthrogryposis. The Turner Scientific and Research Institute for Children's Orthopedics. E-mail: dsbukl@mail.ru.

Буклаев Дмитрий Степанович — к. м. н., заведующий отделением артрогрипоза ФГБУ «НИДОИ им. Г. И. Турнера» Минздрава России. E-mail: dsbukl@mail.ru.

Kostik Mikhail Mikhailovich — MD, PhD, associate professor of the chair pediatrics hospital Saint-Petersburg State Pediatric Medical University.

Костик Михаил Михайлович — к. м. н., доцент кафедры госпитальной педиатрии ГБОУ ВПО «СПбГПМУ» Минздрава России.

Agranovich Olga Evgenievna — MD, PhD, professor, head of the department of arthrogryposis. The Turner Scientific and Research Institute for Children's Orthopedics, Saint-Petersburg, Russian Federation. E-mail: olga_agranovich@yahoo.com.

Агранович Ольга Евгеньевна — д. м. н., руководитель отделения артрогрипоза ФГБУ «НИДОИ им. Г. И. Турнера» Минздрава России. E-mail: olga_agranovich@yahoo.com.

Trofimova Svetlana Ivanovna — MD, Ph.D, research associate of the department of arthrogryposis. The Turner Scientific and Research Institute for Children's Orthopedics. E-mail: trofimova_sv2012@mail.ru.

Трофимова Светлана Ивановна — к. м. н., научный сотрудник отделения артрогрипоза ФГБУ «НИДОИ им. Г. И. Турнера» Минздрава России. E-mail: trofimova_sv2012@mail.ru.