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OPEN TRAUMATIC DISTAL FEMORAL PHYSEAL FRACTURE IN A PEDIATRIC PATIENT WITH TETRASOMY 18p SYNDROME

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Background. Because they are associated with a high energy mechanism, open distal femoral physeal fractures are rare injuries. Appropriate management of these injuries is critical in the pediatric population due to the increased risk of damage to the physis and subsequent growth disturbance and deformity after injury. Pediatric fractures may also represent the sequelae of genetic syndromes (particularly connective tissue disorders), nutritional abnormalities, or conditions that predispose to abnormal bone mineral density.

Clinical case. In this case report, we present the case of a 9-year-old girl with tetrasomy 18p who presented with a grossly displaced right open distal femoral physeal fracture in the setting of isolated, lower extremity trauma. The patient underwent an irrigation and debridement, followed by an open reduction and cross-pinning through the distal femoral epiphysis. After the operation, the patient was made non-weight-bearing in a cast for 4 weeks, and at 6 weeks, the pins were removed. The patient had full range of motion at the 6-month follow-up and then resumed her preinjury level of play at school.

Discussion. Tetrasomy 18p results in inherent muscle weakness that may interfere in the normal soft tissue sleeve of muscles stabilizing long bones. This may result in greater displacement of fractures sustained during trauma, which may lead to a greater rate of vascular injury, physeal injury, and poor overall prognosis. It is imperative for clinicians to be familiar with tetrasomy 18p and its associated orthopedic manifestations.

Conclusion. There is scarce literature on the management of physeal fractures in patients with tetrasomy 18p. In the case described here, we report a good outcome with the standard of care using preoperative and postoperative antibiotics, irrigation and debridement, open fracture fixation, and immobilization in a cast postoperatively. The duration of non-weight-bearing was also increased by 1 week and the pins were removed 1 week later than we would have for patients without any bone or connective tissue disorders.

Keywords: pediatric femur fracture; pediatric distal femur fracture; pediatric physeal fracture; tetrasomy 18p; open pediatric femur fracture.

ОТКРЫТЫЙ МЕТАФИЗАРНЫЙ ПЕРЕЛОМ ДИСТАЛЬНОГО ОТДЕЛА БЕДРЕННОЙ КОСТИ У РЕБЕНКА С СИНДРОМОМ ТЕТРАСОМИИ 18p

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Обоснование. Открытые метафизарные переломы дистального отдела бедренной кости относятся к редким травмам, поскольку являются высокоэнергетическими переломами. Дети, получившие такую травму, нуждаются в своевременном лечении, так как существует повышенный риск повреждения зоны роста с последующим нарушением роста и деформацией кости после травмы. Переломы у детей могут также являться следствиями генетических нарушений (особенно заболеваний соединительных тканей), нарушений питания или состояний, которые вызывают снижение минеральной плотности кости.

Клиническое наблюдение. Рассмотрен клинический случай 9-летней девочки с тетрасомией 18p, которая была доставлена с открытым переломом дистального отдела правой бедренной кости с сильным смещением при изолированной травме нижней конечности. Были проведены туалет и хирургическая обработка раны с после-

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дующей открытой репозицией и перекрестной установкой спиц через дистальный эпифиз бедренной кости. В послеоперационном периоде пациентка находилась в гипсе без опоры на ногу в течение 4 нед. Спицы были удалены через 6 нед. Через 6 мес. наблюдения пациентка свободно передвигалась и снова смогла посещать школу.
Обсуждение. Тетрасомия 18p характеризуется врожденной слабостью мышц, стабилизирующих длинные кости. Это может привести к сильным смещениям при переломах и, следовательно, к учащению сосудистых повреждений, травм зон роста и ухудшению общего прогноза восстановления. Лечащие врачи должны обладать всеми необходимыми знаниями о тетрасомии 18p и связанными с ней ортопедическими нарушениями.
Заключение. В медицинской литературе приведено недостаточно информации о лечении переломов костей у пациентов с тетрасомией 18p. Нами были получены хорошие результаты, соответствующие всем стандартам оказания медицинской помощи, включавшей до- и послеоперационную антибиотикотерапию, туалет и хирургическую обработку раны, фиксацию открытого перелома и наложение гипса после операции. Мы также увеличили продолжительность ношения гипса без опоры на ногу на 1 нед. и удалили спицы на 1 нед. позже по сравнению с ситуацией, при которой признаки заболевания костей или соединительной ткани отсутствуют.

Ключевые слова: перелом бедренной кости у детей; перелом дистального отдела бедренной кости у детей; метафизарный перелом у детей; тетрасомия 18p; открытый перелом бедренной кости у детей.

Background

Tetrasomy 18p is a rare chromosomal disorder with approximately 66 reported cases in literature. It is characterized by the presence of an extra isochromosome 18 with 2 short "p" arms, resulting in a total of 4 copies of the 18p component. The vast majority of tetrasomy 18p cases are due to *de novo* mutations, yet familial inheritance with maternal origin have been reported. The condition has multiple variants but the hallmark orthopaedic features include decreased muscle tone and bone mineral density, scoliosis and kyphosis. [1, 2].

Most patients have developmental delays in motor skills, including sitting, crawling, and walking noted early in childhood [2]. Patients also have non-specific behavior abnormalities, with attention deficit hyperactive disorder and anxiety being commonly reported. Dysmorphic facial features are generally mild and variable. When present they can include low-set ears, small mouth, flat philtrum, thin upper lip, and palatal abnormalities. Once Tetrasomy 18p is suspected, all patients should have an echocardiogram, as up to 30% of patients have structural heart defects. A thorough Gastrointestinal and genitourinary assessment should also be undertaken. While constipation is commonly reported among patients, structural malformations of the gastrointestinal system are not very common. Men are predisposed to the development of hypospadias and crytochordism, while structural changes in the kidneys predisposing to vesicoureteral reflux can occur in both men and women [2].

Despite the association of Tetrasomy 18p to orthopedic deformity, there has been no literature addressing the contribution of musculoskeletal abnormality to injury in the setting of bone trauma [3, 4]. To our knowledge, this is the first case report of Tetrasomy 18p associated with open distal femoral physis fracture. We report on a good outcome for a 9-year old female that underwent a thorough irrigation and debridement, followed by an open reduction and internal fixation.

Description of the case

A 9-year-old female sustained an injury to her right femur after a low-speed sledding accident into an upright basketball pole. The patient presented to the emergency department with gross right knee deformity (figure 1), inability to ambulate, and pain in her right lower extremity.

As an infant, the patient was diagnosed with Tetrasomy 18p. Early in the course of her condition, the patient was noted to have profound neck muscle weakness on clinical exam. She was started on rigorous physical therapy (PT) and was able to independently hold up her head by the age of 3. PT was continued throughout childhood and the patient wore custom foot orthotics until the age of 7, which allowed her to ambulate without assistive devices. She is able to walk and run independently, but tires quickly after a few minutes of activity. She has continued to receive daily physical therapy at school and has no previous history of fractures or related bony trauma.

Imaging revealed a grossly displaced right distal femur fracture without sparing of the physis. (Figure 2). Physical exam was significant for a small poke-hole opening at the posterior aspect of the

thigh. She also had a prominent patella, recurvatum angulation of the limb and obligatory flexion at the knee. The patients mouth was relatively small but she did not have palatal abnormalities or scoliosis. Peripheral sensation and extensor hallicus longus function were intact. A 3+ dorsalis pedis pulse was palpable and dopplerable bilaterally. ABIs in the right and left lower extremities were noted to be 1.2 and 1.1, respectively. The patient's parents were counseled on the possibility of growth arrest. Tetanus vaccination was up to date and 1g IV-Cefazolin was administered prior to transportation to the operating room.

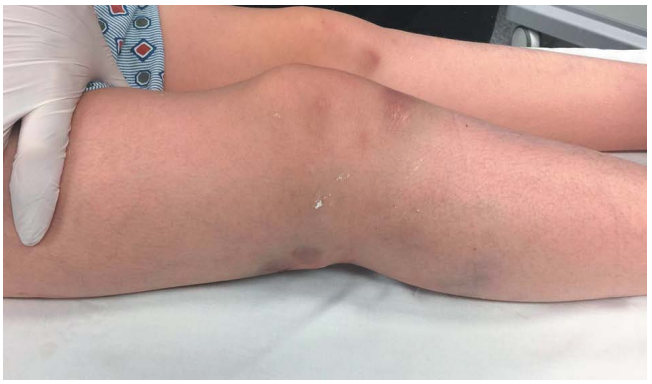

Fig. 1. Picture of the Right lower extremity showing a swollen, grossly deformed right knee

A posterolateral incision at the right distal femur was undertaken. After a thorough irrigation and bridement with 9 L of Normal Saline 0.9%, entrapped periosteum was evacuated and the fracture was reduced. The fracture was fixed in acceptable alignment by cross-pinning through the distal femoral epiphysis. Adequate vascularity was then confirmed by Doppler studies and the wound was closed by primary intention. The patient was placed into a long leg cast and made non-weight bearing. Postoperative plain films revealed an adequate reduction of the distal femur (Figure 3). Since the patient was from out of town and had a 3 hour car ride back home, she remained in the hospital for 2-days to monitor for compartment syndrome, assess for neurovascular compromise, and for completion of 48-hours of post-operative IV-Cefazolin. The patient was discharged and returned to her home state where she had all of follow up visits. The patient remained non-weight bearing for 4-weeks in a cast. The pins were removed at the 6-week mark and by the 6-month follow up visit, the patient had full range of motion and resumed her pre-injury level of play at school. Per the clinic note, final X-rays at 6-months showed maintenance of reduction. The patient was followed clinically for 2 years and no angular deformity or leg length discrepancy was noted.

Discussion

Distal femur fractures through the physeal plate are a rare entity in pediatric trauma. In the upper extremity, physeal fractures can occur after a simple mechanical fall; lower extremity fractures often involve higher energy mechanisms such as motor vehicle accidents [5].

In distal femur physeal fractures, the most common pattern is a Salter-Harris 2, with a fracture line through the physis, exiting via an

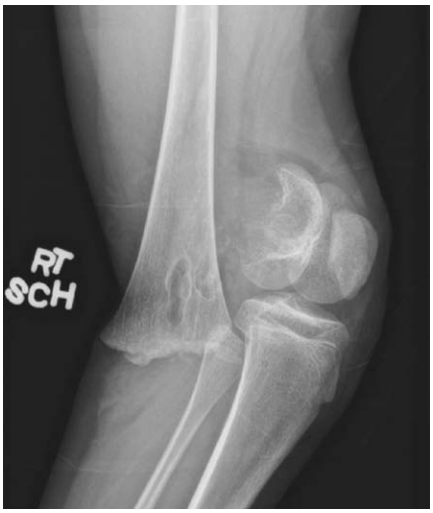

Fig. 2. Post injury Lateral of the Right knee showing a displaced Salter-Harris-2 distal femur fracture

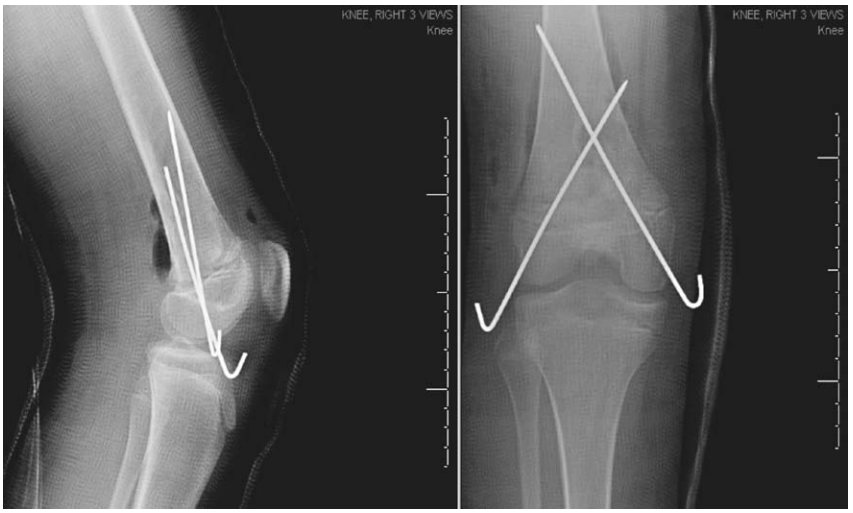

Fig. 3. AP and Lateral of the Right knee following open reduction and internal fixation. Adequate reduction with 2 smooth cross pin fixation through the epiphysis is demonstrated

oblique extension through the metaphysis. Open Salter-Harris 2 fractures of the distal femur are uncommon, given the significant soft tissue envelope. These fractures are commonly associated with high energy mechanisms or multi-extremity trauma. Complications include neurovascular injury, compartment syndrome, infection or physeal arrest following injury and attempted reduction.

Many heritable disorders involve altered bone density or biomechanics, resulting in unique modifications to the above principles of common pediatric fractures. In our case, a relatively low energy mechanism resulted in a significant fracture. This could have been due in part to the patient's decreased bone mineral density. However, the patient's parents do not report any history of previous fractures or skeletal injury. We further postulate that the degree of fracture displacement, to the extent that the proximal femoral fragment pierced the skin of the posterior thigh, is likely due in part to the patient's decreased muscle tone at baseline. As in patients without connective tissue disorders, the normal soft tissue sleeve surrounding the distal femur would be expected to stabilize fracture fragments in such a low energy trauma. Multiple studies have shown that worse outcomes are associated with the degree of initial fracture displacement in these injuries [6, 7]. Therefore, clinicians should be aware of the poor prognosis of distal femur fractures in kids with inherent derangement of bone density and soft tissue.

Vascular injury was a major concern in our case and multiple assessments to ensure adequate vascularity were made throughout the patients care. Popliteal artery injury is as rare as less than 1%, but when present, it is usually associated with hyperextension mechanism [8]. We believe this to be the mechinism in our patient, given the recurvatum angulation of the affected limb and obligatory flexion at the knee. We postulated that the inherent decreased muscle tone associated with Tetrasomy 18p could contribute to the significant displacement and consequently more extensive soft tissue disruption, placing the popliteal artery at higher risk of injury. Based on this rationale, we complementary added Doppler and ABI index to ensure patient's vascular bundle was intact, despite palpable distal pulses.

Distal femoral Physeal fractures, even if minimally displaced, have a high association with growth arrest, with an incidence as high as 58% in some studies [9, 10]. In addition, periosteal entrapment, as in our case has been described as a harbinger for premature physeal arrest [11]. For that reason, we stressed growth arrest as a potential complication, when counseling the parents.

With any pediatric physeal fracture, the full extent of post-injury complications may only present after months or years. Young age favors remodeling but also favors leg length discrepancy. We recommend close long term follow-up to identify any growth arrest of the distal femoral physis. We also extended the non-weight bearing period to 4-weeks and removed the pins at 6-weeks to ensure adequate healing in the patient.

Conclusion

Tetrasomy 18p results in inherent muscle weakness that may interfere in the normal soft tissue sleeve of muscles stabilizing long bones. This may result in greater displacement of fractures sustained during trauma, and hence a greater rate of vascular injury, physeal injury and poor overall prognosis. It is imperative for clinicians to be familiar with Tetrasomy 18p and its associated orthopaedic manifestations. Literature is scarce regarding the management of physeal fractures in patients with Tetrasomy 18p. We report a good outcome with the standard of care utilizing pre and post-operative antibiotics, irrigation and debridement, open fracture fixation and immobilization in a cast post-operatively. We also increased the duration of non-weight bearing by 1-week and removed the pins 1-week later than we would have for patients without any bone or connective tissue disorders.

Additional information

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The patient's informed consent. Informed consent for the patient has been obtained.

Authors involvement

Dr. A.R. Arain is the primary author and was involved with writing, editing and submitting this work.

Drs. M. Moral, K. Desai, C. Adams were also involved in writing and conducting a literature review for the respective topic.

Dr. J. Roberts is the attending and principal investigator of this article who helped with editing.

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