Blount disease in a case of Prader-Willi syndrome: why is it not as prevalent as in obesity without Prader-Willi syndrome?

Susan Dulka, Arabinda Kumar Choudhary, Sosamma Methratta, Kristine Fortuna
Hershey, USA

Case report

Background: **Prader-Willi syndrome (PWS) is a genetic disorder causing multisystem abnormalities with obesity. Obesity is a well established cause of Blount disease.**

Methods: **A 7-year-old girl with PWS presented with genu varum of the left knee with deformity of the proximal medial tibial condyle, which was consistent with Blount disease.**

Results: **A lateral physeal stapling and subsequent osteotomy for the left proximal tibia and fibula were performed with improved anatomical alignment.**

Conclusion: **Blount disease has not been reported in the literature in association with PWS despite excessive obesity.**

World J Pediatr 2013;9(2):182-184

Key words: **Blount disease; genetics; obesity; Prader-Willi syndrome; radiology**

Introduction

Prader-Willi syndrome (PWS) is a genetic disorder due to either deletion of paternally derived chromosome 15q11-13 or maternal disomy 15 (uniparental disomy). The disorder is associated with short stature, hypogonadotropic hypogonadism, mild mental retardation, infantile hypotonia initially causing feeding difficulties and failure to thrive during infancy followed by hyperphagia, and obesity during adolescence.[1] Uncontrollable hyperphagia and reduced activity are the main causes of obesity in patients with PWS.

The etiology of Blount disease in obesity is most likely related to abnormal and asymmetric compressive forces across the proximal tibial physis, causing local growth inhibition and osseous deformities. Most physicians recommend treatment for Blount disease when the tibial metaphyseal-diaphyseal angle (TMDA) is more than 16 degrees. Scott et al[2] outlined the criteria for predicting Blount disease: TMDA greater than or equal to 10 degrees and body mass index (BMI) greater than or equal to 22. Sabharwal et al[3] also observed an increased magnitude of biplanar tibial deformities in patients with the early-onset form of Blount disease.

We present an unusual patient with PWS and obesity, who also presented with Blount disease. To our knowledge, Blount disease has not been reported to be associated with PWS. Obesity is associated with Blount disease and is the likely cause of Blount disease in our patient. The present report highlights this association and describes the causes why Blount disease may be relatively uncommon in PWS despite excessive obesity.

Case report

A 7-year-old girl with PWS initially presented with deformity of the left lower extremity. Initial standing anterior-posterior radiographic images of the pelvis and lower extremities demonstrated genu varum of the left knee with deformity of the proximal medial tibial condyle consistent with Blount disease (Fig. A). Excessive obesity was also noted at the time of diagnosis (Fig. B) and also in multiple prior radiographic studies at 3 years of age. A lateral physeal

Author Affiliations: Department of Radiology (Dulka S, Choudhary AK, Methratta S) and Department of Orthopedics (Fortuna K), Penn State/Milton S. Hershey Medical Center, 500 University Drive, Hershey, PA, 17036, USA

Corresponding Author: Susan Dulka, MD, Department of Radiology, Penn State/Milton S. Hershey Medical Center, 500 University Drive, Hershey, PA, 17036, USA (Tel: 7175310000; Fax: 717 531 5596; Email: sdulka@hmc.psu.edu)

doi: 10.1007/s12519-011-0315-3

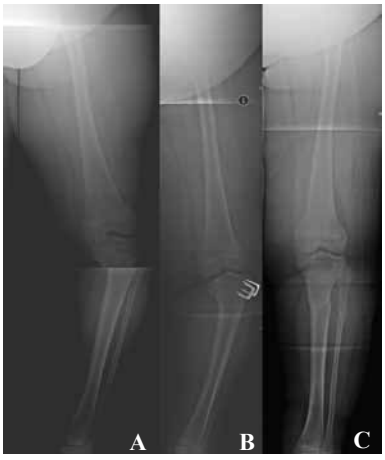

Fig. A: Anterior-posterior view of the left lower extremity demonstrating features of Blount disease on the left side including varus deformity, metaphyseal beaking, irregularity and a wedge shaped medial epiphysis; **B:** Anterior-posterior view of the left lower extremity post left epiphyseal stapling demonstrating residual genu varum of more than 20 degrees; **C:** Anterior-posterior view of the left lower extremity post left osteotomy demonstrating improvement of genu varum.

stapling of the proximal tibia was performed, but it failed to correct the genu varum of over 20 degrees (Fig. B). Subsequently, an osteotomy for the left proximal tibia and fibula was performed. Follow-up images demonstrated improved anatomical alignment of the left genu varus (Fig. C). Her BMI was 45.8 kg/m^2 at presentation and 52.5 kg/m^2 at follow-up.

Discussion

The radiographic findings of Blount disease include medial tibial metaphyseal beaking similar to, but more marked than that seen by physiological bowing. West et al[4] reported the prevalence of orthopedic conditions in PWS children. Among the findings, 47% were related to flat feet, 19% to knock knees, 3% to bowlegs, and 0.2% to slipped capital femoral epiphysis (SCFE). Kroonen et al[5] found that 2 of 29 PWS patients with genu varum were in excess of 5 degrees.

It is striking to find a low incidence of Blount disease in PWS considering the significant relationship of obesity with Blount disease. West et al[4] reported inactive lifestyle, delayed puberty and no tendency to retroversion may be protective for the development of SCFE. Similar arguments may be made for Blount disease. The other potential factor may be gait. Vismara et al[6] found that except for hip joint, the range of motion of knee and ankle joints were significantly reduced in PWS patients compared to both obese and healthy subjects. In relation to knee joint in their study, 63% of PWS patients presented a hyperextended knee in a stance phase, which was likely to be caused by the excessive load that the knee must support. In normal gait the load of the body is supported by the muscle activity of the leg, but in an overweight situation a more pronounced knee extension can reduce the activity of quadriceps and hamstrings. Furthermore, muscular hypotonicity in PWS patients is likely to be the only strategy that allows them to bear their weight while extending the knee. This can be found in a lower percentage of obese patients (35.7%), i.e., the muscles of these patients are able to support the load without extending the knee. The ability of PWS children in sitting, kneeling, standing and walking is delayed as compared to normal children of the same age. These patients therefore develop their typical gait pattern already influenced by obesity.[6] The delayed ambulation in itself may also be protective against Blount disease. Fat distribution is also different in PWS children, who have more fat and less lean tissue compared to those with simple obesity.[7] There was a more feminine fat pattern even in males with PWS, which was thought to be due to delayed sexual development, small gonads and decreased testosterone levels, thus interfering with muscle growth.[7]

In our patient, however, obesity probably did result in Blount disease. Weight management and rehabilitation programs aimed at improving hypotonia and weight reduction as well as stimulating development of motor skills[6] may be an important clinical tool to prevent Blount disease. Understanding of the association of Blount disease with PWS is important for early detection and appropriate management of the disease.

In conclusion, we present an interesting case of PWS with obesity who developed Blount disease. Even in excessive obesity, different factors play a role in developing Blount disease depending on fat distribution, difference in gait, delayed motor skills, relative sedentary life style, increased fat to lean tissue, no metabolic syndrome as compared to obese children without PWS, which probably protects the lower extremity joints from obesity related to PWS. Further multidisciplinary research, including survey of persons with PWS to assess for prevalence of Blount disease, is needed to understand the causes of low incidence of Blount disease in PWS children, which may also prove to be useful to prevent Blount disease in obese children with or without PWS.

Funding: None.
Ethical approval: Not needed.
Competing interest: No competing interest.
Contributors: Dulka S, Choudhary AK and Methratta S helped with the preparation of manuscript and radiographic images.

Fortuna K provided expertise from orthopedic viewpoint and helped with the preparation of the manuscript. Choudhary AK is the guarantor.

References

1 Jones KL. Prader-willi syndrome. In: Jones KL, eds. Smith's recognizable patterns of human malformation, 5th ed. Philadelphia: WB Saunders Company, 1997: 202-205.
2 Scott AC, Kelly CH, Sullivan E. Body mass index as a prognostic factor in development of infantile Blount disease. J Pediatr Orthop 2007;27:921-925.
3 Sabharwal S, Zhao C, McClemens E. Correlation of body mass index and radiographic deformities in children with Blount disease. J Bone Joint Surg Am 2007;89:1275-1283.
4 West LA, Ballock RT. High incidence of hip dysplasia but not slipped capital femoral epiphysis in patients with Prader-Willi syndrome. J Pediatr Orthop 2004;24:565-567.
5 Kroonen LT, Herman M, Pizzutillo PD, Macewen GD. Prader-Willi syndrome: clinical concerns for the orthopaedic surgeon. J Pediatr Orthop 2006;26:673-679.
6 Vismara L, Romei M, Galli M, Montesano A, Baccalaro G, Crivellini M, et al. Clinical implications of gait analysis in the rehabilitation of adult patients with "Prader-Willi" syndrome: a cross-sectional comparative study ("Prader-Willi" syndrome vs matched obese patients and healthy subjects). J Neuroeng Rehabil 2007;4:14.
7 Theodoro MF, Talebizadeh Z, Butler MG. Body composition and fatness patterns in Prader-Willi syndrome: comparison with simple obesity. Obesity (Silver Spring) 2006;14:1685-1690.

Received May 31, 2010
Accepted after revision January 10, 2011