Nail-Patella Syndrome (NPS) is a rare genetic disorder that causes individuals to be born with skeletal deformities and other structural abnormalities. This condition is also referred to as hereditary osteo-onychodysplasia (HOOD), Fong disease, Turner-Kieser syndrome, or Osterreicher-Turner syndrome. One in 50,000 persons worldwide is estimated to have characteristics of the disease. Individuals born with NPS usually demonstrate structural deformities, such as absent or underdeveloped thumb nails and patellae, absent distal interphalangeal (DIP) creases, limited elbow motion, and limited upper arm and leg muscular development.

The purpose of this report is to introduce athletic trainers and therapists to NPS, its most common orthopedic characteristics, and injury prevention considerations for athletic participation. The information presented was derived from an athletic trainer’s experience in working with a 14-year-old male, high school athlete diagnosed with NPS.

A High School Athlete With NPS

The first encounter with the athlete followed the occurrence of a hamstring strain that was sustained while playing on the freshmen soccer team. The athlete reported to the athletic training room complaining of pain in his hamstring muscles that developed after having felt a pop in his right knee during practice. The athlete displayed a noticeable limp, as well as an observable deformity of both knees. The athlete reported the existence of his NPS condition and identified his numerous orthopedic deformities, which included quarter-sized patellae that subluxed each time he flexed his knees. He reported experiencing multiple patellar subluxations on a daily basis (Figures 1-3). As soon as he contracted his quadriceps or actively extended his knee, the patella subluxation would spontaneously reduce. Other manifestations of NPS reported by the athlete included the absence of fingernails on his thumbs and index fingers, the absence of...
distal interphalangeal joints in the fingers due to joint fusion, the inability to extend the elbows fully, and a radial head that protruded about one-half inch with recurrent subluxation from the capitellum (Figures 4-6). Individuals with NPS also have tight muscles in the upper and lower extremities, which may have contributed to this athlete’s hamstring strain.

The athlete’s family history included his mother and two uncles having NPS. Both uncles were very athletic, and one uncle had played professional soccer in Europe, despite his NPS condition. His mother was diagnosed with NPS at age 24, whereas the athlete was diagnosed at birth. He had a sister who did not have any NPS characteristics. Despite the NPS diagnosis, he was encouraged by physicians to participate in various physical activities.

The existence of NPS did not affect the athlete’s treatment for the hamstring strain injury, which resolved without complications within a normal recovery period; however, his predisposition for musculo-tendinous injuries in the upper and lower extremities presented an uncertain prognosis.

A second encounter with this athlete occurred after soccer season, when the athlete was participating as a member of the wrestling team. Having wrestled in junior high school, the athlete was familiar with the demands of this sport. His small stature placed him in the lightest weight class (under 105 lbs). All of his wrestling coaches were aware of the NPS condition and previously had discussions with the athlete about risks associated with his participation in the sport.

During the wrestling season, the athlete reported knee pain that was described as having had a gradual onset. This visit occurred late in the wrestling season and was thought to be due to excessive kneeling. He was treated for symptom relief and provided with additional knee padding. His participation in practice sessions was also limited, in order to ensure that he would be able to compete in an upcoming match.

A final encounter during the same academic year occurred during one of the athlete’s late-season wrestling matches, during which an opponent pulled his arm behind his back. Because his elbow extension was
limited by NPS, the result was a mild elbow hyperextension sprain. He subsequently recovered from the injury and was able to finish his wrestling season without further incident.

**General Appearance**

Individuals with NPS usually have a lean body composition and have difficulty in increasing weight or building muscle. A deficiency in muscle mass may be noticeable, especially in the upper arms and legs. Male-pattern baldness (i.e., a high and broad forehead) is also commonly seen at a young age. Other physical manifestations of NPS include deformities of the knees and fingernails and the inability to completely extend the elbows.

**Fingernails and Hands**

Fingernail abnormalities are the most common deformity, which are found in 80-90% of individuals affected by NPS. The fingernails of the thumbs and index fingers are most commonly affected, and the severity of fingernail deformity tends to lessen for each successive finger in the ulnar direction (i.e., 3rd, 4th, and 5th). The nails will appear brittle, cracked, or underdeveloped, and tend to be more deformed on the ulnar side of the nail. The most predictive clinical sign for the existence of NPS is triangular-shaped lunulae on the thumbs (i.e., the normally crescent-shaped areas of nail bed growth at the bases of the nails). Toenails can have the same deformities but are less commonly affected.

Another signature characteristic of NPS is the absence of creases in the skin of the DIP joints. Individuals with this condition will not have a normal amount of DIP flexion (Figure 7). The loss of flexion tends to be most severe in the index finger, with the severity lessening toward the 5th digit.

The athletic trainer can assist in preventing damage to the athlete’s fingernails by routinely inspecting the nail beds for infection and by recommending use of gloves for protection of the nails (if sport rules permit their use). Batting gloves should be worn when playing baseball, and receiving gloves should be worn when playing football. Soft cloth gloves should be worn for cross country or soccer. Even though the athlete’s hands are rarely used in these sports, the possibility exists that the athlete’s fingernails could be avulsed if caught on another athlete’s uniform.

An athlete whose fingernails are absent should be treated the same as an athlete who has had a nail removed. The area should be covered and kept as clean as possible, and the nail bed should be routinely checked for infection. If infection occurs, immediate referral to a physician is essential for proper treatment.

**Knees**

Abnormalities of the knee, usually affecting the patellae, are present in 98% of reported NPS cases. The patellae may have obvious deformities, such as being small or irregularly square-shaped, or they may be completely absent. Size and shape of the patellae will vary between the left and right knees, and the presence or absence of a patella in one knee may not correspond to that of the opposite knee. These patellar abnormalities are associated with frequent subluxations.

Knee deformities predispose the athlete with NPS to a greater risk for development of degenerative arthritis. Other possible abnormalities associated with knee function include osteochondritis dissecans, tight hamstrings, and the absence of an anterior cruciate ligament. These conditions may lead the athlete to report knee symptoms such as a locking or clicking, feeling unstable, and giving way and may increase susceptibility to ligamentous, meniscal, and musculotendinous injuries. Athletic trainers and therapists should encourage athletes with NPS to wear custom-made knee braces during athletic participation.

**Elbows**

Deformities of the elbows tend to be associated with a loss of extension, pronation, and supination (Figures 8-10), which relate to antecubital pterygia (i.e., skin webbing). Elbow abnormalities, such as a prominent medial epicondyle and hypoplasia of the lateral epicondyle and capitellum, often contribute to recurrent conditions.
dislocations of the radial head. The combination of these abnormalities with the force imposed by falling on an outstretched arm commonly cause posterior elbow dislocations in athletes with NPS. Athletic trainers should consider recommending the use of custom-made elbow braces to restrain elbow hyperextension during participation in contact sports.

**Summary**

The likelihood that an athlete with NPS has not been diagnosed as having the condition is low, but the possibility exists that an athletic trainer may have to care for an athlete with NPS when participating as a member of an athletic team. Although NPS is relatively rare, athletic trainers should be familiar with the orthopedic abnormalities associated with the condition. Because athletes with NPS possess normal cognitive and emotional function and a desire to participate in competitive athletics, athletic trainers should take a proactive approach to reduce the incidence and/or severity of injuries that could result from the orthopedic abnormalities associated with the condition.

**References**


*Jennifer M. Plos* and *Renee L. Polubinsky* are Athletic Training Education Program faculty with the Department of Kinesiology at Western Illinois University in Macomb.

*Joe McKown* was a graduate assistant, Certified Athletic Trainer for Western Illinois University and Macomb High School when this article was written.