

# Nail Patella Syndrome

## Common Occupational Therapy Practices Shed Light on a Rare Genetic Disorder

In July 2002, Nail Patella Syndrome Worldwide held its fifth International Symposium in Pittsburgh. For two days, patients, families and clinicians had an opportunity to explore and discuss every aspect of this rare and unusual condition. Nail Patella Syndrome (NPS) is a genetic disorder characterized by nail and skeletal deformities. It occurs in about one in 50,000 people, and it is inherited in an autosomal dominant matter, which means the risk of transmission is 50 percent per pregnancy, regardless of gender. In 12 percent of the cases, the disease is the result of a new or spontaneous mutation. It is found throughout the world in all ethnic groups.

NPS manifests itself in a wide range of symptoms and severity. The most noticeable characteristic is the absence or underdevelopment of fingernails, especially the thumb nail. Skeletal anomalies also may include underdevelopment of the patella (kneecaps), iliac horns or spurs on the pelvic bones, permanently dislocated elbows which affect the mobility and rotation of the wrists, twisted legs or club feet, and curvature of the spine. It appears that several other medical conditions are closely associated with NPS, including kidney problems, glaucoma and digestive disorders.

UPMC's Dr. Adele Towers has a son with NPS and was instrumental in bringing the conference to Pittsburgh. "The International Symposium is an excellent opportunity for NPS patients and their families to gather lots of information," says Towers. "This is most often a condition that runs in families, and so the conference has a certain 'family reunion' atmosphere."

As an internist specializing in geriatrics, Towers is attuned to the important role that occupational therapy plays in measuring and improving function. So as plans were underway for the 2002 conference, she noticed the glaring absence of occupational therapists in the broad mix of medical participants.

Towers contacted Dr. Margo Holm, Professor, Department of Occupational Therapy, and asked if she could help conference participants begin to look at the syndrome's affect on function. As Holm began to study the clinical features of the syndrome, she selected tools that would provide data on how elbow and knee involvement –

present in over 70 percent of NPS patients – affects function. She then turned to Ketki Desai, an occupational therapy doctoral candidate, to implement the first-ever study of how Nail Patella Syndrome impacts functional participation in everyday activities.

Desai – with the help of Emily Eckel and Dr. Tamara Mills – collected data at the Pittsburgh conference. The study involved 32 women and 13 men, ranging from 12 to 69 years of age. All of them had either elbow or knee involvement, with 67 percent having both. Data collection began with a Health Assessment Questionnaire – 21 questions which delved into patients' level of mobility, self-care and domestic life.

"This group consistently rated themselves with a very low level of difficulty in performing everyday tasks," says Desai.

"We were most likely dealing with a representative group of the most highly functional NPS patients – because they were able to travel to the conference." However, many participants indicated some level of difficulty with bending, lifting and wrist turning activities.

The next step in this study involved the Keitel test – a performance-based assessment of the range of motion of upper and lower extremities. Participants were assessed on their ability to flex and extend knees, rise from a chair, and position their hands in various ways.

Desai wanted to take a closer look at hand function, so she also used the Jebsen Taylor Hand Test, a performance-based assessment of supinating rotation of the wrist. Participants were timed as they turned cards, simulating page-turning.

"The number one complaint among the patients we worked with was the elbow," notes Holm. "For some with NPS, cutting meat, buttoning a shirt and opening a jar are quite challenging. And just imagine trying to go to a drive-thru ATM machine without the ability to rotate the elbow and wrist joints."



Holm describes the occupational therapy strategies that can be used for intervention with NPS patients. "It is clear that many of the participants in our study already employ adaptive strategies to assist them with everyday tasks. However, an occupational therapist can offer compensatory approaches such as toilet seat lifts and kitchen devices as well as restorative approaches such as exercise or splinting. And some basic educational approaches can help NPS patients really make the most of their physical functionality."

Dr. Iain McIntosh, a geneticist at Johns Hopkins University who identified the gene for NPS, is the leading researcher on this condition, also known as Iliac Horn Syndrome, Hereditary Onycho Osteodysplasia (HOOD), Fong's Disease and Turner-Kieser Syndrome. McIntosh has invited Desai to share her findings with colleagues around the world and to continue and expand the occupational therapy study in conjunction with the ongoing work at Johns Hopkins.

The results of the data collected in Pittsburgh were presented at the 6th International Symposium on Nail Patella Syndrome in June 2003 in St. Louis, Missouri, and later at the First Medical Conference for NPS-UK in August 2003 in Chester, England.

"Ultimately, the more information we gather about NPS, the more we can help patients get the kind of medical services they need," notes Towers. "The folks who came back to the 2003 conference in St. Louis were very excited by the results of the 2002 occupational therapy study. They told us that no one had ever asked them about functionality before – and no one knows better than they do what an impact NPS has on day to day life."

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