PRADER-WILLI SYNDROME ASSOCIATION

An Open-Label Pilot Study of N-Acetylcysteine for Skin-Picking in Prader–Willi Syndrome

by Jennifer L. Miller, M.D., M.S., Dept of Pediatrics-Endocrinology, University of Florida, Gainsville, Florida, and Moris

Skin-picking can result in serious and potentially life-threatening infections. Recent evidence suggests that the excitatory neurotransmitter glutamate is dysregulated in obsessive- compulsive behaviors, and modulation of the glutaminergic pathway may decrease compulsive behaviors, such as recurrent hair pulling or skin-picking behaviors.

Nacetylcysteine (NAC), a derivative of the amino acid cysteine, is thought to act either via modulation of NMDA glutamate receptors or by increasing glutathione in pilot studies. Thirty-five individuals with confirmed PWS (ages 5–39 years, 23 females/12 males) and skin-picking behavior for more than 1 year were treated with Nacetylcysteine (Pharma-NAC) at a dose of 450–1,200 mg/day. Skin-picking symptoms and open lesions were assessed after 12 weeks of treatment by counting and measuring lesions before and after the medication.

All 35 individuals had improvement in skin-picking behaviors. Ten (29%) individuals (six males and four females) did not have complete resolution of skin-picking behavior, but had significant reduction in the number of active lesions. Longer-term, placebo-controlled trials are needed to further assess the potential benefit of this treatment.

(**Post note:** Nacetylcysteine can be bought without a prescription and there are various brands. Dr. Miller states Pharma-NAC is a good brand because it does not degrade. It should not be started without first talking to your child's doctor, making sure there are no drug interactions with any of his/her prescribed medications, and getting a starting dose recommendation.)

Am J Med Genet Part A 9999:1–4. Drs. Miller & Angulo are members of PWSA (USA) Clinical Advisory Board