

A multidisciplinary approach to the provision of custom equipment for children and adults with Lesch Nyhan Disease

Alice Wintergold, Chailey Clinical Services, Sussex

Summary

Lesch Nyhan Disease (LND) is a rare genetic disease, with those affected typically having dystonia, choreoathetosis and self-injurious behaviour. This presentation will explore Chailey's experiences of working together as a multidisciplinary team in order to assess, design and provide custom made equipment for children and adults with LND.

Aims & Objectives

To present Chailey's multidisciplinary approach to the provision of equipment through the Activities of Daily Living Clinic.

To provide information about the condition with a focus on the self-harming issues often associated with LND.

To present the key components of the clinic.

To present the outcomes of the clinic that have been found to be successful.

Background

LND is a rare genetic disease with normally affects boys (McCarthy et al 2011). Those affected typically have a neurological disability, self-injurious behaviour and an overproduction of uric acid (Jathar et al 2016). Self-injurious behaviour includes lip and finger biting (Gonzales et al. 2018), hyperextension of the neck and banging of the head. Carers may also be injured.

Managing the self-injury is challenging, and medication has not been found to be routinely effective. Local services often struggle to meet the needs of those affected and their families. Commercially available equipment is often not suitable due to the opportunities for injury. Local teams can find it difficult to provide appropriate advice on provision of equipment to help manage this behaviour.

Chailey Clinical Services provides an outpatient clinic that can be accessed nationally. The clinic offers the opportunity to access a multidisciplinary team (MDT) of therapists, doctors, clinical scientist and rehabilitation engineers. The team members have experience of advising on management of this condition, and consider a range of options in terms of specialist equipment and therapeutic/medical advice. Liaison with the local team and family is essential prior to the clinic in order to prepare materials, assessment equipment and team member availability.

At the clinic the MDT holistically consider the clients requirements for a range of activities such as accessing the curriculum, communication, powered mobility and personal care. In particular assessment and/or custom-made equipment can be designed, manufactured and trialled in order to find unique solutions to each client's requirements.

During the clinic week the MDT build a relationship with the client and family. An important part of this is demonstrating that the team understand the condition and will work together to keep the client safe and secure. Those with LND need to test new situations and equipment to make sure they are safe, and the testing phase is important in the successful introduction of new equipment. The feeling of security then leads to a reduction in the self-injurious behaviour.

Discussion

Providing equipment for those with LND is complex due to the combination of the requirement for supportive equipment in conjunction with protection from harm. The focus is on the client with considerations of the expectations.

Using an MDT approach enables the holistic needs of both the client and family to be considered throughout the assessment, therefore providing a solution that is likely to be more robust and longer lasting to account for potential changes. The MDT approach enables the clinic to be flexible and adapt as required.

The clinic environment, time and facilities enable the assessment equipment to be set up and trialled. Equipment may include modifications to standard products or custom-made items being manufactured for the assessment. Equipment often enables a safe position in sitting, lying, standing and for personal care. Clients may also benefit from the use of mitts and equipment to assist with manual handling. The provision of appropriate equipment for clients with LND reduces self-injurious behaviour and increases participation in activities.

References

- González, C , Díaz V, Riesco F and Chávez H. (2018). Oral self-mutilation in Lesch-Nyhan Syndrome. Case Report. Rev Chil Pediatr. 89(1) pp86-91.
- Jathar P, Panse A M, Jathar M, and Gawali P N. (2016). Lesch-Nyhan Syndrome: Disorder of Self-mutilating Behavior. Int J Clin Pediatr Dent. 9(2) pp139–142.
- McCarthy GT, Green EM, Ogunbona O, Simmonds HA, Fairbanks L, Pountney T, Bryant E.(2011). A population study of Lesch-Nyhan disease in the UK. Dev Med Child Neurol. 53(1) pp34-9.

Email: alice.wintergold@nhs.net