Will there be an impact on the child's education?

Education will continue to be important for the child or young person and family and there will be many aspects that require consideration and significant assistance from those around them. Specialist Visual Impairment services exist to support both children and young people and the education professionals working with them and these will play an important role in facilitating the process.

The BDFA offers support to any family member, friend, professional or organisation involved in caring for a child or young person with CLN3 disease or any other form of NCL throughout the UK. We provide informed guidance and assistance as well as seeking to increase awareness of the disease and facilitate research to identify potential therapies and ultimately a cure.

We organise workshops, conferences and are able to arrange connections with other affected families. The BDFA also coordinates a Small Grants Scheme that can provide assistance for a range of needs. The BDFA has a Support and Advocacy Partner who is able to assist with many of the issues highlighted in this document and can discuss each of them in greater detail and on a more personal basis.

The BDFA family folder can also provide further specific information on CLN3 disease. The folder is free for all families and available to professionals at a cost of £25.

Where can I get additional information and support?

Please contact the BDFA Family Support and Advocacy Partner via our Freephone Helpline: 0800 046 9832 or email support@bdfa-uk.org.uk for further information and to order a copy of the family folder.

The BDFA can provide information on a number of local and national organisations that are also able to offer various forms of support and information that will be relevant to families. It may also be appropriate to refer a family to be made to local children’s hospice service, as this can offer an additional experienced and skilled source of holistic support.

What are the chances of inheriting CLN3 disease?

Since the first genes causing NCL were identified in 1995, over 400 mutations in 14 different genes have been described that cause the various forms of NCL. Our cells contain thousands of genes that are lined up along chromosomes. Human cells contain 23 pairs of chromosomes (46 in total). Most genes control the manufacture of at least one protein. These proteins have different functions and can result in the NCLs caused by abnormal genes, which are unable to produce the required proteins. As a result, the cells do not work properly and this leads to the development of symptoms associated with these diseases.

What specifically causes CLN3 disease?

The gene called CLN3 was discovered in 1995 and lies on chromosome 16. This gene codes for a transmembrane protein and mutations (mistakes) in the CLN3 gene cause deficiencies that result in abnormal storage of proteins and lipids (fats) in neurons (nerve cells) and other cells. The most common condition that results from a deletion of part of the gene, which is present in 85-95% of all CLN3 disease. The cells then cannot function properly and this leads to the development of the symptoms associated with CLN3 disease.

How are NCLs inherited?

Most forms of NCL are inherited as ‘autosomal recessive’ disorders. This is one of several ways that a genetic disorder can be passed down through families. An autosomal recessive disorder means that both copies of the gene are abnormal (one inherited from each parent) and neither parent is affected. The disease does not depend on the sex of an individual.

What are the real implications for the family?

As the disease progresses, specialist equipment and aids will become necessary and this is another area where the family will need help. Items are likely to be focused on addressing challenges associated with living with a visual impairment, though will ultimately include specialist seating, wheelchairs, bathing and toileting aids, hoisting equipment, and a specialist bath or shower. Professionals will play a key role in ensuring that these and other items are provided in a timely manner following proper assessment of the individual child and young person. It is likely that changes will be needed in the home environment to enable the family to appropriately care for a child or young person with CLN3 disease. These may initially include adaptations to promote the independence for living with a visual impairment e.g. specialist lighting, tactile labeling, introducing contrasting colours for objects and areas, as well as installing suitable floor surfaces. In the latter stages of the disease it may be necessary to install ramps, widen doorways or invest in a purpose-built wet room with a specialist bath or shower, whilst there are various other aspects that will require consideration.

Will there be an impact on the child's education?

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The information in this document is provided on the understanding that it is intended for general information purposes only, therefore the Batten Disease Family Association (BDFA) accepts no responsibility or liability for any other form of use, nor for circumstances arising from any unintended or misconstrued use. The BDFA have made every effort to ensure that the information provided is appropriate and accurate at the time of publication. All decisions pertaining to care and treatment of an individual child should be managed, in conjunction with parents/legal guardians, by qualified professionals working for the appropriate health, education and social services.

Some of the information contained in this leaflet is based upon chapters in “The Neuronal Ceroid Lipofuscinoses (Batten Disease) 2nd Edition” by Mole, Williams & Goebel (Eds), Oxford University Press 2011 and is used with permission.
Children appear to be healthy and develop normally for the first few years of life. The first sign of the disease usually presents as a gradual loss of vision between 4 and 7 years of age, which may first be noticed in a nursery or school environment. Each child’s level of vision will change rapidly over a 6 - 12 month period initially, however it is likely that some awareness of colour along with variations between light and dark will be retained until later. By the end of their attendance at primary school, children tend to begin showing some difficulties with concentration, short-term memory and learning. Many are still able to attend a mainstream school though may require additional learning support in the classroom. Changes in behavioural patterns can become apparent at various stages of the disease and may prove particularly challenging to address and manage.

- **Sleep disturbance can be a problem in CLN3 disease.** This is best managed through promoting various activities such as walking, swimming, cycling and riding. Regular psychotherapy and other similar input should be utilised as the disease progresses with a multi-disciplinary team assessing the needs of the child or young person.

**What is Sleep Disturbance?**

- **Insomnia**
- **Night terrors**
- **Sleepwalking**
- **Night sweats**
- **Enuresis**
- **Sleep apnoea**
- **Nocturnal leg muscle twitches**

Despite these issues, many children and young people are able to get a good night's sleep. It is important to try to be consistent and have a regular routine. This can help them relax and get into a sleep pattern. Other things that may help are to sleep in a comfortable and quiet environment, avoid screens before bed, and try to get up at the same time every day.

**Drug treatment of sleep problems**

Options include

- Antipsychotics such as risperidone
- Antidepressants such as sertraline
- Anxiety medicine such as clonazepam

**What are the symptoms and how does the disease progress?**

**CLN3 disease** is a rare genetic condition affecting children and young people. The disease progresses in stages, and the symptoms and challenges can vary from child to child, even within the same family. The symptoms can include:

- **Visual symptoms** (e.g. difficulty seeing certain colors, loss of vision)
- **Seizures**
- **Muscle stiffness**
- **Behavioral changes**
- **Sleep disturbances**
- **Gastrointestinal issues**

**During the teenage years, children and young people slowly become less able and increasingly dependent, yet the course of the disease varies rapidly depending on fatigue, underlying illness etc).**

**Deterioration motor skills e.g. problems with balance and walking can be first noticed when promoting various activities such as walking, swimming, cycling and riding. Regular psychotherapy and other similar input should be utilised as the disease progresses with a multi-disciplinary team assessing the needs of the child or young person.**

**What is Deterioration in Motor Skills?**

- **Difficulty walking or balance**
- **Muscle stiffness**
- **Fatigue**
- **Ataxia**

**It may also be possible that older unaffected siblings also carry the disease as carriers of the disease and may want to understand how CLN3 disease may affect their family choices when they are older.**

**Is support available to families?**

As soon as possible following a diagnosis of CLN3 disease, families should be offered support from various professionals attached to their local health, social, educational, visual impairment services and the CLN3 Support and Advisory Team. Ideally a “Team Around the Child or Young Person” will be formed, with one of the professionals appointed as a Keyworker for the family.

The child or young person’s needs should be discussed with the keyworker and successful strategies agreed. The team will work to ensure that the child or young person and family receive the ongoing care and support they need and that their choices are taken into account.

A child or young person and their family’s needs will inevitably change as the disease progresses. It is therefore important that any child will be an individualised plan of care and support is essential.

**How can families manage the financial challenges?**

Caring for a child or young person with CLN3 disease will bring additional financial challenges. It is vital that families are well informed about the full level of economic assistance available and the support that they are entitled to. They may need help and guidance in accessing benefits and other sources of assistance. The professionals and services supporting the family should provide advice and guidance. The BDFA can also support families with these issues in various ways, the Small Grants Scheme being one particular example.