the child or young person. It remains probable that many parents
will continue to need guidance, understanding and support when
they are now better classified according to the gene identified as
the cause e.g. CLN3 disease, juvenile and CLN1 disease, infantile.

Where can I get additional information and support?

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
information, guidance and assistance as well as seeking to
increase awareness of the disease and facilitate future research
to identify potential therapies and ultimately a cure.

We organise conferences, workshops and are able to arrange
connections with other affected families. The BDFA also
coordinates a Small Grants Scheme that can provide assistance
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of support and information that will be relevant to families. It may
also be appropriate for a referral to be made to a local children’s
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any unintended or unsanctioned use. The BDFA has 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 or young person should be managed, in
conjunction with a doctor and/or therapist, by qualified professionals working for the appropriate health, educational 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 Mele, Williams & Goebel (Eds), Oxford University Press 2011 and is used with permission.

Edited by Harriet Lammens (QVH), Dr Christine Caven (MRCP) Matt Hobbs (RF Nurse) and Professor Paul Clowen, Batten Disease
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What are the chances of inheriting CLN3 disease?

The diagnosis of CLN3 disease is usually made by tests on blood
samples, though was more commonly known as Juvenile Batten Disease.

They will have a 50% (1 in 2) chance of inheriting one abnormal gene, which
would make them a carrier who is unaffected by the disease. There is a 25% (1 in 4)
chance of the child being born with two normal genes and therefore being non-
affected (not a carrier).

When it is known that both parents are carriers of the abnormal gene, we refer
to being there being a 2 in 3 chance of a child being a carrier, once it
is established that they are unaffected by the disease.

How is it diagnosed?

Children or young people will probably have been seen by a
paediatric and paediatric ophthalmologist due to a progressive
loss of vision. A number of investigations will often have been undertaken to look for the cause of the presenting visual impairment.

The diagnosis of CLN3 disease is usually made by tests on blood
samples, though a skin biopsy may also be necessary.

How common is it?

Approximately 3 - 4 children or young people are diagnosed
with CLN3 disease each year in the UK. We estimate there are
currently between 30 - 40 affected children and young people
in the UK. Children and young people have been diagnosed
with this condition in many countries and from a variety of ethnic
backgrounds.

what are the practical implications for the family?

As the illness progresses, specialist equipment and aids will
become necessary and this is another area where the family will
need help. Items are initially likely to be focused in 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 bed/mattress.

Professionals will play a key role in ensuring that these aids items are provided in a timely manner following proper
assessment of the individual child’s or young person’s needs.

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 particular
adaptations to promote independence for living with a visual
impairment e.g. specialist lighting, tackle labelling, 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, wider 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.

Are there any alternative names?

CLN3 disease, juvenile may also be referred to as juvenile CLN3
disease. It has previously been called Spiekmeyer-Sjogren-Vogt
disease and Juvenile Neurocervic Lipofuscinosis (JNCL);
though was more commonly known as Juvenile Batten Disease.

What can Feasibility can be caused by?

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 mutation
is 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.

CLN3 Disease is inherited as an autosomal recessive disorder,
which means that both chromosomes carry mutations in the
CLN3 gene. Therefore both biological parents of a child or young
person with this diagnosis, will be carriers of the disease but
physically unaffected by it.

A child born to parents who both carry the autosomal recessive
mutation in the CLN3 gene, has a 25% (1 in 4) chance of
inheriting the abnormal malfunctioning genes from both parents
and developing CLN3 disease.

With any pregnancy, the probability of a child inheriting one
or both genes from their parents is the same each time, irrespective
of any sibling’s status.

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with CLN3 disease each year in the UK. We estimate there are
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in the UK. Children and young people have been diagnosed
with this condition in many countries and from a variety of ethnic
backgrounds.

What are Neuronal Ceroid Lipofuscinoses (NCLs)?

These refer to several different genetic life-limiting neurodegenerative diseases that share similar features. Although
the different forms of NCL are sometimes described according to the age of the child or young person at the onset of the
disease, they are now better classified according to the gene identified as
the cause e.g. CLN3 disease, juvenile and CLN1 disease, infantile.

What causes NCL?

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 disease. 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 include enzymes which act to speed up molecular
chemical reactions. The NCLs are 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?

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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”
diseases. This is one of several ways that a genetic disorder
can pass on through families. An autosomal recessive disorder
means that both chromosomes of the gene are abnormal (one
inherited from each parent) with neither working properly. The
result is that the body cannot produce the protein that is present
in normal cells and so is unable to carry out its normal functions.
In many cases this results in the development of the symptoms
associated with the disorder.

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

Children appear to be healthy and developing 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. Most children will become blind by the end of their attendance at primary school. Children appear to be healthy and developing 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. Most children will become blind by the end of their attendance at primary school.

**Are there any treatments?**

Currently there is no cure for CLN3 disease and therefore specialist symptom management and therapy is essential to assist in the daily life of the child and young adult and to support the family. Holistic support for parents, siblings and wider family members is extremely important throughout their journey.

Epileptic seizures start on average at age 10. The most common type are tonic-clonic seizures where limbs can jerk and loss of consciousness occur. Other types of seizure include partial seizures where only one part of the body is affected by abnormal movement and altered consciousness may not be very obvious. A consultant neurologist’s guidance is essential for managing seizures. The monitoring of effectiveness of medication, seizure control and checking for side-effects needs to be done as a collaboration between families of affected children or young people and their neurologist often working with other professionals such as GPs and specialist nurses.

Drug treatment of seizures depends on their severity and frequency. Anti-epileptic medications such as sodium valproate and lamotrigine, sometimes needed in combination, are often the first treatments introduced. Other medications used in addition include clobazam, clonazepam, levetiracetam, phenobarbital and phenytoin.

The anti-epileptic drugs carbamazepine and gabapentin have been found to be of no use in CLN3 disease.

In the later stages of CLN3 disease combinations of anti-epileptic drugs may be needed. For clusters of seizures drugs from the benzodiazepine group such as midazolam, diazepam and lorazepam given either orally or via a gastric tube can be very effective. Paraldehyde is a drug given rectally in cases where clusters of seizures are resistant to other medications.

Emotional, behavioural and psychological difficulties are common in all stages of CLN3 disease with the potential for psychotic episodes to appear in the later phases of the disease. These include restlessness, anxiety, panic attacks, aggressive behaviour, hallucinations, delusions and depression. Familiar supportive environments that are peaceful and structured can make a significant difference in managing these challenges along with a focus on promoting self-esteem and flexibly adapting activities to each individual’s abilities (acknowledging that these can sometimes vary rapidly depending on fatigue, underlying illness etc.).

Sometimes medication is of use in dealing with behavioural problems, hallucinations and agitation. Drugs from the benzodiazepine family such as diazepam or klonopin can be helpful. Other medications that have been used successfully are risperidone, olanzapine and chlorpromazine. All of these types of medication can cause significant difference in managing these challenges along with a focus on promoting self-esteem and flexibly adapting activities to each individual’s abilities (acknowledging that these can sometimes vary rapidly depending on fatigue, underlying illness etc.).

Sleep disturbance can be a problem in CLN3 disease. This is best managed by attention to a sleep routine and making every effort to help with strategies that help a child or young person relax. If medication is needed then options include melatonin or one of the drugs from the benzodiazepine family.

Deteriorating motor skills e.g. problems with balance and walking can be first addressed through various activities such as walking, swimming, cycling and riding. Regular physiotherapy and other similar input should be utilised as the disease progresses with a focus on maintaining mobility for as long as possible and, although there can be great variation in each individual, there will ultimately be a need for mobility aids and other specialist equipment for which Occupational Therapy input is invaluable.

At some stage a child or young person with CLN3 disease is likely to develop difficulties with chewing and swallowing. It can become a challenge to maintain adequate nutrition and hydration which can lead to weight loss and problems giving medication. Symptoms of reflux can occur and may lead to distress and pain so causing behavioural disturbances. There may be difficulty with swallowing saliva leading to excessive dribbling. Difficulties in swallowing can sometimes cause choking and put a person at increased risk of chest problems.

Professionals including dieticians, speech and language therapists, psychologists, occupational therapists, social workers, occupational therapists, dieticians, psychologists and general practitioners should be involved to help with these problems. Medications such as acid suppressants and prokinetics can be used to help with swallowing problems.

Research into possible methods for treating the disease is ongoing with various theoretical approaches being considered and investigated.

Current strategies being examined include modulating the immune system, blocking classes of glutamate receptors, and targeting dysphagia. However, there are many scientific questions yet to be answered and all are currently at very early stages of development.

For updates and information regarding developments in research please visit the BDFA website: www.bdfa-uk.org.uk or contact the BDFA Scientific Officer via 0800 046 9832 email: research@bdfa-uk.org.uk

**What research is being done?**

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**What are the genetic considerations?**

The age that CLN3 disease is usually diagnosed in a child or young person means that some families will have younger siblings who may be affected but have not displayed any symptoms.

It may also be possible that older unaffected siblings are carriers of the disease and may want to understand how CLN3 disease may affect their family choices when they are older. When only one parent is a carrier of the disease, then there is a 50% (1 in 2) chance that any child will be an unaffected carrier.

If parents are considering having additional children, they can access specialist advice and support from their local clinical genetics service following a referral from their GP. Prenatal testing may be possible in the early stages of any future pregnancy.

**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 the local, regional or national specialist services and the BDFA Support & Advocacy Partner. 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 parents and assessed by the team. The team will work together 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. As such, it is often helpful if a clear process for regular reviewed reviews is identified and that a system is established for enabling additional reviews as and when they are deemed necessary. As the rate or pattern of the progression of the disease for each child or young person remains uncertain, an individualised plan of care and support is essential.

The BDFA is able to provide various forms of holistic support and can be contacted via 0800 046 9832

email: support@bdfa-uk.org.uk

**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 all the possible financial assistance available and the support that they are entitled to. They may well 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.

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