



Infantile Batten Disease

To get a blue disability vehicle badge a young person would normally have to qualify for high level Disability Living allowance. The High Rate Mobility Component of your DLA can sometimes be used to lease or hire a vehicle through the **Motability Vehicle Scheme**, see www.motability.co.uk 0845 456 4566.

Mobility allowance can sometimes be used for the **Motability scheme** to fund a vehicle. Just because a young person has a particular disease does not mean that the application will automatically be successful, it is the level of disability that is important - not the diagnosis. Each case is judged on its merits. As the disease progresses, the level of disability increases, it is important not to "give up" if unsuccessful - keep trying. There is an appeals process if the claim is unsuccessful.

If a parent has to care for the child for more than 16 hours per day and the child receives DLA then the parent may be entitled to **Invalid Carers Allowance**.

Further advice can be obtained from the Citizens' Advice Bureau, The Benefits Agency or a social worker.

Equipment

As the illness progresses, various kinds of equipment and aids become needed:-

- Car seats
- Pushchairs / wheelchairs
- Specialist mattress
- Seating for in the home
- Protective headwear for when the child is still mobile but falling frequently
- Postural splints for the torso or limbs
- Feeding related - feed pumps and consumables (feed, containers and syringes)

- Suction for pharyngeal secretions
- Incontinence pads ["nappies"]
- Hoisting equipment or even lifts
- Bath aids

The ease or difficulty of obtaining these items and resources varies greatly around the country, but all should be available without any cost to the family. Very expensive equipment provision may be means tested to a degree.

Education

In the early stages of the illness, often before the correct diagnosis has been made, it is already clear that help will be needed with schooling. Most families find this very difficult and upsetting. At a time when the realisation is beginning to sink in, that their son or daughter isn't going to be "OK" and will need a lot of extra help to make progress at school, parents often find many bureaucratic obstacles to be overcome in order to get the help they feel their child needs. In trying to get through the process of "statementing", parents need guidance, understanding and support. "Statementing" is the process where the child becomes the subject of a **Statement of Special Educational Needs**. This is a formal document drawn up by the local education authority, based on advice from the health professional and other professionals involved - usually at least a community paediatrician and an educational psychologist. It describes the child's educational needs and makes recommendations for the appropriate educational provision and allocation of resources. Unfortunately the resources provided often fall short of the resources needed. Adequate schooling is a field where considerable experience exists within BDFFA.

Introduction and Terminology THE IDEA BEHIND PRODUCING THIS LEAFLET IS TO PROVIDE FAMILIES AND THE PEOPLE AROUND THEM WITH AN OVERVIEW OF THE ILLNESS. OUR UNDERSTANDING OF WHAT CAUSES THE ILLNESS IS GROWING RAPIDLY AND THIS LEAFLET TRIES TO EXPLAIN THE SCIENCE IN AN EASY TO COMPREHEND WAY.

The terminology is confusing. This is mainly because what we call Batten Disease (named after the British neurologist who described the condition in 1903) is in fact several different genetic illnesses each caused by a different gene defect. They do however have many similar features. The different types of "Batten Disease" are often classified according to the age of the child when the illness begins, so there are infantile, late infantile and juvenile types of Batten disease for example, with the illness seeming to start at around 1, 3 and 7 years of age respectively.

As our understanding at the level of molecular genetics has increased, things have become more complex. The Batten Disease group of illnesses now tends to be referred to as the **Neuronal Ceroid Lipofuscinoses (NCL)** this term describes the abnormal appearance of affected cells as seen down a microscope. It is now clear that there are some less common "variant" types - some of these are quite rare and only occur in significant numbers in certain parts of the world.

Molecular Genetics

Our chromosomes contain tens of thousands of genes, most of which control the production of proteins. These proteins have various functions and include enzymes which act to speed up molecular chemical reactions. We know that the NCLs are caused

by abnormal genes, and that the required proteins are not made properly. Eight gene defects are known so far, and they cause the different varieties of NCL - these are known as CLN 1 to 8. The 8 gene defects are all different and so are the abnormal proteins. All somehow end up with a similar disturbance in cell function which results in similar clinical symptoms. All the proteins involved seem to be involved with the function of the lysosome. This is a tiny part of a cell, and its function is to break down and recycle certain chemicals in the cell. When this doesn't work properly, there is a build up of substances that can't be broken down properly. This 'build up' becomes toxic to the cell and eventually the cell dies.

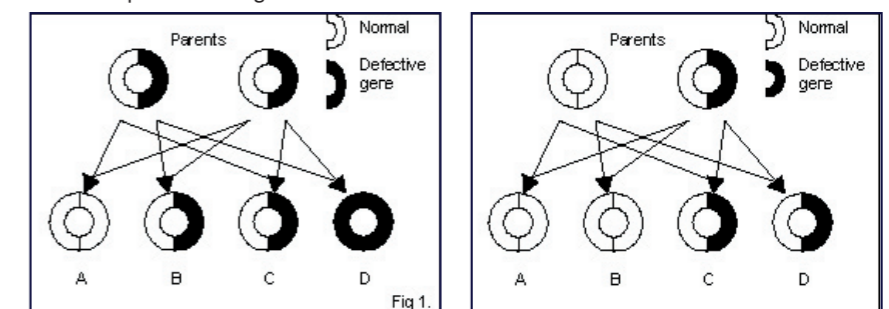
How are NCLs inherited?

Apart from some very rare types, the NCLs are inherited in an "Autosomal Recessive" fashion. *Autosomal* means that the abnormal gene is carried on the normal chromosomes as opposed to the "sex" chromosomes. This means that the illness has nothing to do with gender. (Some illnesses are carried on the sex chromosomes and are said to be sex-linked and principally affect either males or females - such as colour blindness or haemophilia). *Recessive* means that the abnormal gene does not lead to an affected individual if he or she has a normal gene as well. We each have a pair of genes - one passed down from each parent. If both genes are normal the

individual does not have the disease. If a person has one normal gene and one abnormal gene, he or she does not have the disease (but is a carrier and could pass the gene on to his or her child). If a person has two abnormal genes, he or she has or will get the disease.

The diagram below (Fig 1) tries to illustrate the various ways parental genes come together at conception when both parents are carriers of the disease. Child A has two normal genes and does not have the disease. Child D has two abnormal genes and so does have the disease. Children B and C have one normal and one abnormal gene - they do not have the illness but will be carriers. For each pregnancy, there is a one in four (25%) chance that the baby will be affected; a one in four chance that the baby will have two normal genes and be healthy, and a two in four (50%) chance that the baby will be healthy but a carrier. Two out of three healthy children will be carriers.

If only one of the parents is a carrier as in Fig 2, no children have the disease but 50% will be carriers (on average) This explains how the gene can be passed down family trees from our ancestors for many generations without showing until **bad luck** brings two carriers together. When this happens (as shown above, in Fig.1) each child of this couple will have a one in four chance of being affected by Batten Disease.



This leaflet is for basic information only; any decisions regarding an individual child should be done through the appropriate medical services. Some of the information in this leaflet is based on the chapter on Classic Infantile NCL by Dr P. Santavuori et al in The Neuronal Ceroid Lipofuscinoses, Goebel, Mole & Lake (Eds), IOS Press 1999 and is used with permission.

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Heather House, Heather Drive, Tadley, Hampshire RG26 4QR Tel: 07914 060742
www.bdfauk.org / www.bdfauk.co.uk E-mail: info@bdfauk.co.uk

