



BATTEN DISEASE FAMILY ASSOCIATION

Bringing light to Batten's

OUR MISSION STATEMENT

***THE BDFA'S VISION IS TO
BRING LIGHT TO BATTEN DISEASE BY BEING THE
CENTRAL POINT OF EXCELLENCE IN THE UK FOR
SUPPORTING AFFECTED FAMILIES AND TO FACILITATE
RESEARCH INTO THE DISEASE***

OUR OBJECTIVE

Our objective is to be the central point of excellence in the UK for the support, guidance and networking of affected families and professionals as well as to increase awareness and facilitate research into Batten Disease. To do this we have already established the foundation support principles for the charity and we are now expanding to offer more informed guidance and support as well as actively increasing awareness of the disease and facilitation of research and clinical trials.

THE NEED

Batten Disease is the main paediatric neurodegenerative disorder in the UK. It affects 1:30,000 births and there are approx 200 affected children/ young adults in the UK. They are born apparently healthy, develop epilepsy, and lose their sight, speech, mental and motor abilities before dying between the ages of 5-30. There are 3 main types of Batten Disease, affecting children at different ages depending on which defective gene has been inherited, traditionally called Infantile, Late Infantile and Juvenile Batten Disease. There is also a congenital form and other variants of the disease. There is much research into all types, but no cure at present.

Having Batten Disease profoundly changes the child's and their family's life forever. The journey from even before diagnosis to death is a forever-changing picture of needs as the disease progresses. Unlike other major diseases where there is a huge awareness of the support, therapies and guidance mechanisms needed for the child and family, the families of Batten Children are left isolated and numbed as most professionals do not understand the course of the disease and the needs the child and families have. It is therefore essential that the families and professionals have a place to turn to for that support and guidance. This is the principle aim for the Batten Disease Family Association so that the affected children, young adults and families do not feel isolated and are given the tools to be able to continue to live life and reach their maximum potential.

The other need is for a UK Batten Disease organisation to take responsibility for guiding the future direction and facilitation of the management of Batten Disease in the Health, Education and Social Services and to fully support and promote UK research/ clinical trials into the all types of the Disease as well as link into the network of other Batten organisations in the world.

BACKGROUND TO THE BDFa

The BDFa was formed in 1998 and gained charity status in 2001. It has expanded to 200 members, with another 70 professionals linked through our professional organisation, the Batten Disease Interest Group formed in 2005. The BDFa also supports many families and professionals who are not members, UK and worldwide. The BDFa is the only UK Batten Disease organisation offering this support and guidance. It provides a wealth of information already and advice for families, professionals and the public and a national networking opportunity to share experiences.

To help develop the charity, the BDFa in 2003 was effective in securing Jeans for Genes Campaign funding to identify 'Best ways in coping with the disease' and to create a Clinical Assessment Tool, to help predict the course of the disease to help plan needs accordingly and as a monitor for any clinical trials in the future. These are now being completed and in the next 2 years need to be translated into working practices and communicated to families, professionals and researchers. With this grant and others, the BDFa has so far supported funding of over £60,000 into research.

In 2006, the charity enrolled its first Development Officer in order to facilitate the delivery of the BDFa's aims and has used this role initially to establish the foundation stones for the charity to further expand the support already given.

THE FUTURE VISION

Over the next 5 years the BDFa will continue to optimise the direct support it gives affected families. The BDFa now plans to expand into more indirect support and focus on making sure that the professionals around the affected families have the correct tools to help them. The BDFa also plans to actively increase the awareness of Batten disease generally, contributing to national decisions on management of the disease and increase facilitating and help co-ordinating UK and worldwide clinical and research developments.

Over the last 10 years, research has developed significantly and much more is understood about the disease with many more gene variants of the disease identified. For most Batten Disease types, research is at or will come to a point of needing to trial different therapies over the course of the next decade, both in the laboratories and clinically. The BDFa plans to contribute significantly to the course of these developments, particularly in the UK. At the same time, the UK public services are all looking now at ways in which they can help manage the disease and need that national voice, which the BDFa plans to develop.

SUMMARY

The BDFa is already the only dedicated UK Batten Disease charity supporting the families and research. It now aims to become a stronger UK voice for Batten Disease in order to communicate and implement members' wants and needs and influence UK decisions on the management of Batten Disease and facilitate any research or clinical trials.

Ultimately the BDFa wants a cure for each of the types of Batten Disease, but there are many stages to manage before that is achieved. By having a UK organisation committed to the health and wellbeing of affected children and young adults through the support, understanding and education of families, carers and professionals and also optimising Batten Disease research and development efforts into supporting and curing, the BDFa will be helping move effectively towards achieving that ultimate aim.