

**Juvenile Batten's disease**  
**Thoughts on its Traditional Chinese Medicine diagnosis and some**  
**observations of working with a patient with this degenerative disease**

This essay will look at some thoughts on the traditional Chinese medicine diagnosis, as well as some observations and reflective practice aspects of dealing with a patient with Juvenile Batten's disease. The essay will be written as follows. Firstly, a description of the disease from a western perspective with the problems of taking a case and the signs and symptoms involved. Secondly, some thoughts as to how this might be interpreted by traditional Chinese medicine and thirdly my approach and some observations and reflective aspects of this.

According to Stedman's medical dictionary (1995) Batten's disease is a form of cerebral sphingolipidosis. Cerebral sphingolipidosis is "any one of a group of inherited diseases characterised by failure to thrive, hypertonicity, progressive spastic paralysis, loss of vision and occurrence of blindness, usually with macular degeneration and optic atrophy, convulsions and mental deterioration".

Sphingolipidosis is the old name for lipofuscinosis.

According to Medhelp (1998) Batten's disease was named after a British paediatrician called Batten who first described the disease in 1903. It can also be called Spielmeier-Voigt-Sjogren-Batten disease and is one of a group of diseases called Neuronal Ceroid-Lipofuscinoses (NCL).

The main type is the juvenile form but there are 3 other forms, all of which are progressive neurodegenerative diseases.

SeeAbility (1998) reports that the infantile form has its onset between 6 months and 2 years, which is rapidly progressing with seizures, dementia, blindness and a severe loss of neurones. Death normally occurs in mid childhood. According to Holton (1997) it is due to an enzyme defect in Palmitoyl-Protein Thioesterase which causes the build up of lipofuscins and is due to a defect on chromosome 1. The late infantile form has an onset between 2 to 4 years, leading to seizures, blindness, loss of muscle co-ordination, mental deterioration and dementia. Death is around 8 to 12 years of age. The enzyme and chromosome abnormality is the same as for the infantile form. The juvenile form has its onset between 5 and 12 years. It causes developmental regression with vision loss, seizures, loss of motor ability and dementia. Death may occur from late teens to early thirties. The enzyme defect is unknown at present but the storage substance is again lipofuscins and the chromosome defect occurs on chromosome 16 on gene CLN3. The adult form is called Kufs or Party's disease and usually appears before the age of 40. It has milder symptoms than the others and it also progresses slower. It does not cause blindness but does shorten life expectancy.

The childhood forms are due to autosomal recessive disorders, which means that they can only occur if the child inherits two copies of the defective gene, one from each parent. Therefore if each parent is a carrier then the child faces a one in four chance of inheriting two defective genes. The adult form can also be due to an autosomal recessive inheritance but also less commonly, by an autosomally dominant inheritance (all people who inherit a single copy of the gene defect will get the disease).

The symptoms of Batten's disease are due to the build up of lipopigments (lipids and pigments) in the body's tissues (brain, eye, skin, muscle etc.). The pigment part is due to a yellow green colour when viewed under an ultra violet light microscope, which is used to identify the disease in skin samples. The pigments look like half moons or fingerprints in the cells. The build up may be due to over production of the lipopigments or as mentioned above the failure of the breakdown of the products by

defective enzymes. According to Stein (1994) lysosomes engulf and degrade both endogenous and exogenous materials and may also be part of the metabolising of low density lipoproteins and some hormones by endocytosis and subsequent incorporations. Patients with lipid storage diseases will accumulate high levels of lipids in the tissues and these tend to cause derangements in the central nervous system development.

There are five stages of the disease: -

- 1) Visual impairment and retinal changes
- 2) After about 2 years there is the onset of generalised seizures and occasional shock like contractions of a set of muscles which may vary in regularity, symmetry and synchrony (myoclonus).
- 3) Gradual intellectual deterioration, slow movements, cerebral ataxia (inability to co-ordinate muscle activity during voluntary movement) and intention tremor.
- 4) Onset of severe dementia, muscles are wasted, although tendon reflexes are lively and the plantar reflexes are extensor.
- 5) The patient finally lies curled up, blind and speechless with occasional dystonic postures.

A search was done of the ARRC data base which did not produce any references in complementary medicine at all, and of the other western papers produced, these were of a biochemical or genetics origin. The following discussion is from the writer's observations on a patient, Davina with the juvenile form.

I have known Davina for about 9 years now, as our next door neighbour. But because of her illness we did not see her often. Her sister, Jenny I know well, as she is a great friend of my daughters. Jenny is five years older than my daughter is, but because of her sisters illness we often felt that she missed out on things and so we would often invite her along when we went out and she has become like part of our family. It was only since starting to practice that her mother, Linda, asked me if I could help Davina at all.

Davina was having hallucinations particularly at around 7pm and also at about 3am. Davina is on many drugs to control her epilepsy and there was not much the doctor could do apart from with consultation with the consultant, upping her drugs to very high levels which Linda was not keen on. Linda was also very tired as she has to give Davina drugs every 6 hours and this is tiring enough without being woken at 3 am. My first reaction was to be very cautious as I was afraid of disturbing anything that might make things worse and cause a major fit. My problem also was that she could not communicate with you, although her close carers could often tell whether she agreed or not. So how was I to take a case history?

My second problem was of the signs and symptoms that I could see what were the true signs and symptoms and what were the drugs.

The drugs that she was on were as follows: -

Epilim – Sodium valproate – side affects gastric irritation, ataxia, tremor, weight gain, oedema, impaired hepatic function. Affects the SP, Damp forming, Blood reducing. Clobazam – side affects drowsiness, fatigue, dizziness, muscle hypotonia, irritability, and hypersalivation in infants. Possible affects in TCM terms - Damp forming, Yin, stagnating, Qi reducing?

Lamotrigine – side affects fever, malaise, blurred vision, dizziness, insomnia, ataxia, irritability, tremor, and agitation. Possible affects in TCM terms - causes Yin deficiency and stagnation of Qi?

Sinemet – Co-caldopa (mix of carbidopa and levodopa) – anti parkinsonism drug – side affects nausea, insomnia, agitation, dizziness, tachycardia, arrhythmia, involuntary movements, flushing and sweating – Possible affects in TCM terms -Yin reducing, moves Qi?

Thioridazine – anti agitation – side effects hypotension, affects vision, drowsiness, pallor, nightmares, insomnia, depression. Possible affects in TCM terms – Qi stagnation, heat affecting the HE?

Frumil – Co-amilofruse- side effects hypotension, increased calcium excretion, GI disturbances, confusion, dry mouth, rashes. Possible affects in TCM terms – Yin deficiency, Wind heat disturbances?

Chlormethiazole – used in fitting phases – side effects nasal and conjunctival irritation, headache, tachycardia, apnoea, hypotension.

Vitamin E – for coating the brain cells

Voltarol – diclofenac sodium – side effects fluid retention, headache, dizziness, vertigo, renal and liver problems. Possible affects in TCM terms – Damp forming and Yin?

This added up to a great cocktail and clouded the true signs and symptoms substantially.

I took her pulse, which was wiry and slippery on the left first and second positions, and deep on the third position, on the right it was slippery in the first and second position and again deep in the third. I did my best at looking at her tongue, which she could not stick out, and if you were lucky you got a glimpse of it. It was red with a thick white coat. I gathered as much information that her mother could supply.

So how can this disease be interpreted into Traditional Chinese Medicine? Putting together the signs and symptoms of the different stages covered earlier in this essay along with Davina's signs and symptoms this is my interpretation.

Firstly, the disease seems to be from a hereditary origin, which as the Kidney Jing is responsible for this, would seem to be the root of the problem. The failure to develop mentally would also point towards the Kidneys, as they are responsible for producing marrow and nourishing the brain. If there is Kidney deficiency there may be poor sight and memory amongst other symptoms. The actual development of the reproductive aspect of the patients with the juvenile form seems to develop as normal along the seven year (in women) and eight-year cycles (in men). But although the physical aspects seem to develop normally, the mental aspects of maturity do not run parallel with this. This part of the Kidneys power seems to be partly reduced, and of course as the patient does not live to middle age it might be said that the Kidney energy is not sufficient to maintain the individual through the whole of their maturation and then their decline. Again this appears to be due to the Kidney Jing or essence. According to Maciocia (1989) Kidney essence is the material basis for both Yin and Yang, even though Kidney essence deficiency is part of Kidney Yin Xu.

Kidney essence is the organic basis for the transformation of Kidney Yin into Kidney Qi by the warming and evaporation of the Yang. Therefore the Kidneys are the root of both Yin and Yang and this will be important a little later in this essay.

Secondly, the visual impairment aspect, as it is to do with the eyes one would think that the Liver has a role to play here but as already explained the Kidney essence nourishes the eyes and may be involved also. From the Liver aspect, could the build up of the lipopigments be due to the fact that the Liver is not playing a proper part in the free flow of Qi and Blood, so that build up of waste materials occurs? This seems to be unlikely, as the disease is so severe to be just a case of stagnation. Perhaps the nourishing of the eyes with Liver Blood / Yin may have a part to play but again the leap from blurred vision to blindness seems to be too great. According to Clavey (1995) the essence from the 5 Yin and 6 Yang organs flows upwards to irrigate the eyes and so problems with the eyes may be due to more than one organ disharmony and this I think is much more likely.

Thirdly the seizures and muscular contractions have several aspects. The Liver controls the sinews. If the Liver Blood is deficient, the sinews will lack moisture and nourishment which may cause contractions, spasms or impaired extensor or flexor movements, numbness of the limbs, muscle cramps, tremors, tetany or lack of strength of the limbs. The Spleen also has a role here for it is probably not

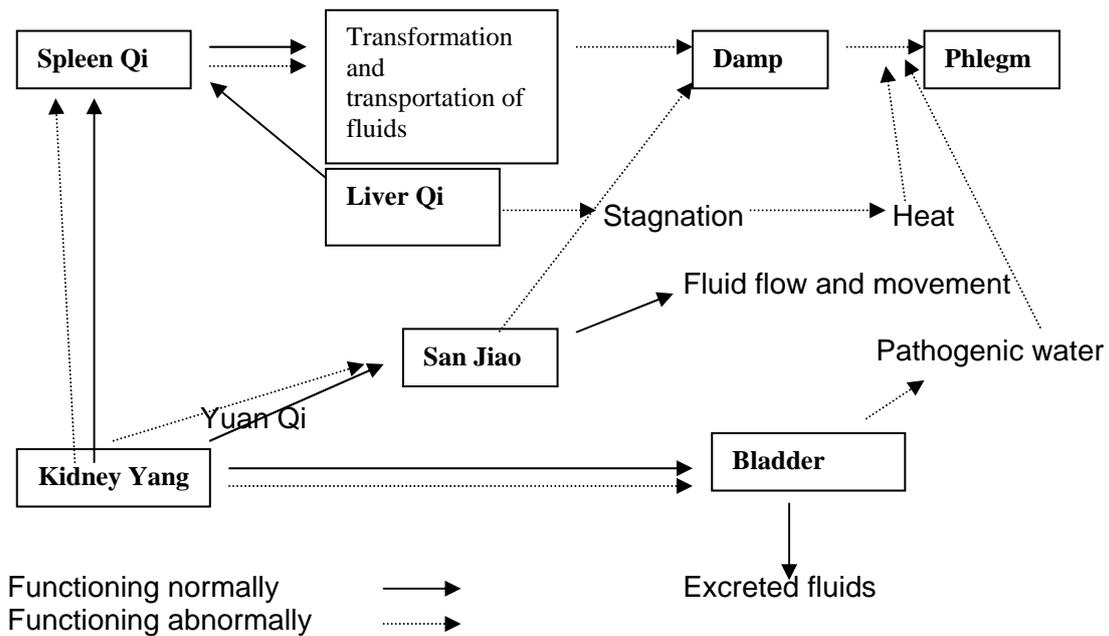
performing its transporting and transforming of food and fluids properly so that the Blood may be made in the Heart with help from Yuan Qi from the Kidneys. The lack of Liver Blood may also be part of the formation of Liver Wind, which can cause the eyeball to turn upward and have nystagmus (which is often seen in these patients). This also leads us on to explaining the convulsions and seizures. This is due to internal Wind, which is again associated with the Liver. It can also be due to constitutional Yin deficiency which allows the Liver Yang to rise which causes internal Wind. The connection between the Kidneys and the Liver can be seen from a five-element point of view in the fact that water is not nourishing wood. So if the Kidneys are deficient then there is the possibility of the development of Liver problems such as Blood and Yin deficiency which may then cause the formation of internal Liver Wind.

Observing the tongue and pulse, there is also Phlegm involved in Batten's disease. The Phlegm is carried up to the head with the Wind to cause the convulsions, and epileptic attacks but can also obstruct the Heart orifices and mist the mind causing epilepsy. Davina also drooled which may indicate Phlegm. The Phlegm aspect will now be explained in much greater detail, as the writer believes this to be a key role in this disease.

Phlegm formation may be by several different mechanisms, the various pathways of which will now be discussed along with how and to what extent they may contribute to JBD.

If the Spleen and the Stomach are in disharmony and the SP is not transforming and transporting fluids efficiently then the fluids will condense to eventually form Phlegm. This Phlegm is often deposited in the Lungs (the Lungs store Phlegm) and if not removed by the Lungs can back up on the SP. If Liver Qi is not assisting the SP by carrying the fluids around the body they will congeal and form Damp, which will eventually form Phlegm. Liver Qi stagnation also produces heat which helps to condense fluids into Phlegm. If Kidney Yang fails to support the SP then the SP transformation and transportation mechanisms will be impaired and it can also deprive the San Jiao of the warming Yuan Qi. This is necessary for fluid movement and for Qi transformation of fluids. Kidney Yang also steams the fluids held in the Bladder and if this is failing this leads to pathogenic water going upwards to the SP which will lead to Phlegm formation. In Juvenile Batten's disease the SP aspect does not appear to be the original problem area but as time progresses the Kidney deficiency will affect the SP. There are also other aetiological factors, which will make the SP Xu worse. Because of the patient being unable to feed themselves and problems with the patient fitting during eating tube feeding is employed. In order to use tube feeding, processed foods are used and so this may not help the problem. Phlegm can also be formed in the Lungs due to the LU failing to disperse and descend fluids to the Bladder where fluids are transformed and then excreted. LU Yin Xu, SP Qi Xu and invasion of pathogens can also cause this. In juvenile Batten's disease this does not appear to be a main contributory factor although, due to the generalised weakened state of the individual they are very susceptible to respiratory problems.

The Kidneys are the root of Yin and Yang, and also receive and store the Jing essence from transformation of food and fluids which, is then re-supplied when required. Long standing Phlegm conditions have some Kidney involvement where weak Kidney Yang or imbalance of Kidney Yin and Yang underlies Phlegm production, hence the phrase the Kidneys are the root of Phlegm. The Kidneys produce a breakdown in fluid transportation or gradual lack of support in the function of the ST or SP or impaired Kidney vitality may cause worsening of an already existing Phlegm condition.



**Diagram to show some of the secondary pathways to Phlegm formation in Juvenile Battens Disease**

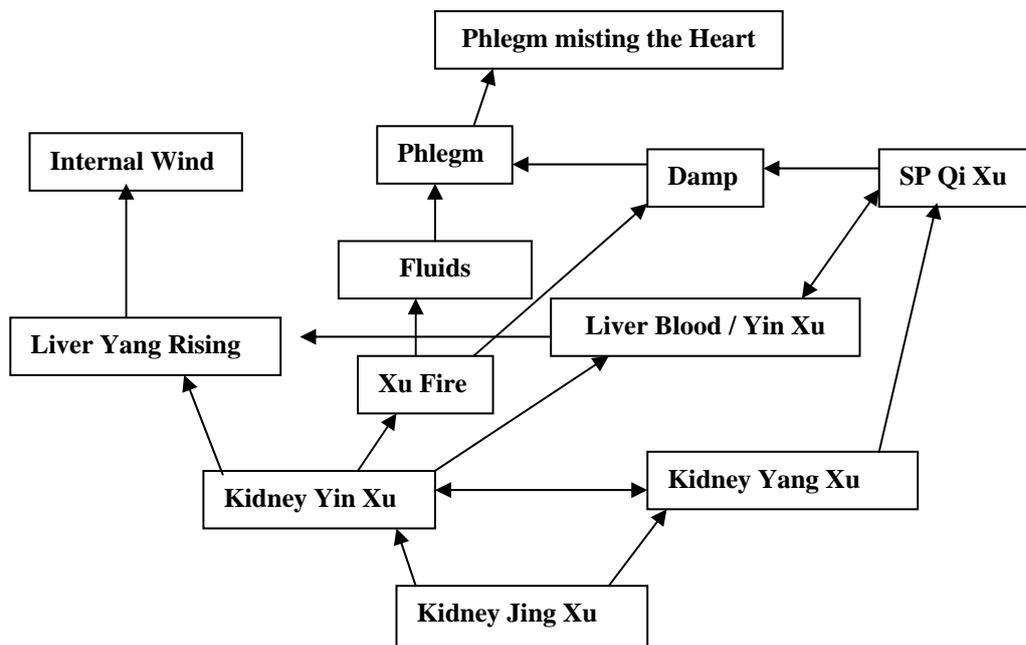
Kidney deficiency can also produce pathogenic fire. This fire does not affect Qi transformation and transportation but dries fluids to form Phlegm, internal Wind, insufficient Yin and extravasated Blood. If there is constitutional deficiency of Yin this may lead to Xu Fire. According to Clavey (1995) "Phlegm is merely fire with form, fire is merely formless Phlegm". It might be remembered that Davina's tongue had a red body and a thick greasy white coat. The pulse was also slippery and wiry. This indicates the probable presence of Phlegm and heat.

The San Jiao also may have some involvement due to the lack of the warming yuan Qi of the Kidney; the SJ may not be able to control the Qi, which goes to warm and nourish the muscles and the flesh and which also makes the skin firm. The muscles and flesh of the JBD patients tend to be very soft and pallid. The lack of Qi and fluid circulation also may cause a blockage in the upper jiao and so more heat may be produced which causes congealing of the fluids into Phlegm.

As Clavey (1995) states the Chinese saying that Phlegm is formed through "the Four Qi and the One Pathogen". The four Qi being LU Qi Xu, SP Qi Xu, KID (Yang) Qi Xu, SJ yuan Qi and the pathogen being the overacting of pathogenic Fire. In the case of the JBD the initial formation of the Phlegm probably came from the constitutional Kidney deficiency and the production of Xu Fire. This led to the production of Phlegm, which has led to Phlegm misting the Heart, accounting for the mental confusion, lethargic stupor and aphasia. This constitutional Kidney deficiency also is unable to hold down Liver Yang rising, which produces Liver Wind, which causes the epileptic fits.

Having gleaned as much information as possible, I decided to treat the symptoms that were causing the most problems, which were the hallucinations.

Davina at this stage was 17 years old and although she was technically a minor I felt from an ethical point of view that I should try to get consent from her but this was extremely difficult. I had to rely upon her mother and carers to give me feedback on this initially. As I got to know Davina I would be able to sense when she was happy and when she was having a bad day.



**Pathology Diagram of Juvenile Battens Disease**

I also consulted another acupuncturist before doing anything for guidance. We decided to keep it very simple and just try one point Heart 8. We chose Heart 8 for its ability to clear heat in the Heart. This I hoped would reduce the dreams / hallucinations which was due to the Shen not being able to rest in the Heart due to the heat. This I knew was a painful point but also knew that if I were quick it would minimise the discomfort. So with the help of her Mum and another carer this is what I did. She showed no reaction to the needle but she had difficulty controlling her limbs and so help was needed to stop her rolling over her arm or clenching her fist. I left it a few days and then went in again, she had seemed to calm down and the hallucinations were less frequent. This I continued through the summer each week (8 weeks) and the 3 am calls stopped and the hallucinations were much more infrequent. As I got to know Davina I found she had a very wicked sense of humour, I would tell her about some thing that had gone wrong either to me or to my daughter and she would after a slight delay start to giggle. As September started she started school again and the 7pm hallucinations and little fits returned. Heart 8 did not seem to help much so I tried a slight difference in prescription as I felt a little more confident, this time I used SI3, BL62 and P5. The reasoning behind this was the combination of SI3 and BL62 opens the Du channel and subdues internal Wind and so is good for convulsions and epilepsy. SI3 also calms the Mind and BL62 is good for daytime epilepsy as it clears interior Wind and it benefits the eyes. P5 was used for resolving Phlegm in the Heart and calms the Mind and so can be used for epilepsy. According to Deadman (1998) it can be used for manic raving as if seeing ghosts and as Davina seemed to be seeing something and becoming very agitated I thought this point might help. This was not easy to do and was not very successful as the first time she seemed a little shaky and this increased the tremors so I took the needles out and held Kidney 1, which calmed the tremors. I decided to return two days later. This time Heart 8 plus holding Kidney 1 seemed to help and things got a bit better. Kidney 1 subdues Wind and empty heat and so is good for epilepsy. It

calms the Mind and as it has a strong sinking action it brings down Liver Wind from the head. It also tonifies Yin and subdues empty heat. Other points that might be useful but were not tried at this stage are ST40 for its ability to calm the Mind when misted by Phlegm, P6 for calming the Mind and is used for epilepsy. We have carried on like this since. Sometimes she has not been well enough, and recently she got a chest infection and ended up in hospital. But she has such spirit and determination to fight on and she is now back at home.

As I treat her I have had encouragement from her family and carers as even a little improvement to Davina's quality of life not only affects Davina but also her family. The family is accepting in the fact that she may not live many more months or years and takes each day at a time. I am always in admiration of their courage but I know that they will have great pain to come when she does go. It brings it home to me sometimes how fragile life is and how the human spirit can win through. I do find it difficult to accept that one day she will not be there. Regardless of the difficulties, if I can help even a little to make her life a little better then it is well worthwhile. I can never hope to 'cure her' but this has been a very useful learning for me that even the impossible should be tackled as quality of life can be improved.

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