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BATTEN DISEASE- A RARE CASE REPORT WITH AYURVEDIC PERSPECTIVE

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ABSTRACT

Batten disease, also known as neuronal ceroid lipofuscinoses (NCL), refers to a group of rare neurodegenerative disorders that usually begin in childhood. The disorder affects a cell's ability to break down lipofuscin, a waste product that accumulates in cells over time. Brain cells are particularly vulnerable. The buildup of proteins and lipids (fats) causes cells to gradually lose function and die.[1] Prevalence of Battens disease in worldwide is similar to one for every 100,000.[2] NCLs are having symptoms like seizures, dementia, visual loss, cerebral atrophy. Eventually children with this disease become blind, wheelchair bind, bedridden, unable to communicate and loss all cognitive functions. There is no effective treatment is available to halt in progression of this disease. Here a case report of Battens disease based on ayurvedic understanding is explained. Ayurvedic explanation regarding nervous system disorders are found under the umbrella of Vatavyadhi.

KEYWORDS: Battens disease, NCL, Seizure, Cerebral atrophy, Dementia, Vatavyadhi.

BACKGROUND

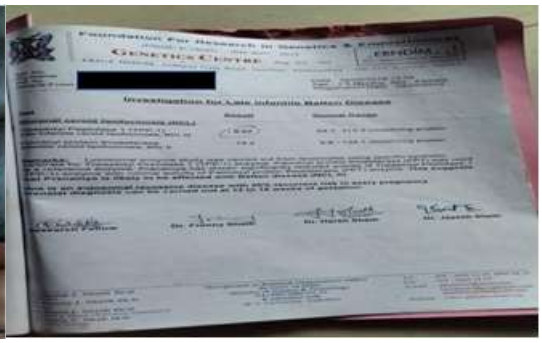
Fredric Batten was a British pediatrician who described this disease in year 1903. Most forms of Batten disease/NCLs usually begin during childhood. Incidence of this disease in India is very few. Children of these forms during infantile or late infantile shows symptoms less than 1year of age. Batten disease is an inherited genetic disorder which is caused due to defect in one among the13 genes. The mutated genes in battens disease does not produce proper amount of proteins which are important for lysosomal function. Each gene provides defective information for a specific protein. Due to lack of functional protein causes abnormal buildup of junk material in lysosomes and abnormal buildup of lipofuscin which occurs lysosomal breakdown of lipids.

Children with all forms of batten disease have shorter life expectancy, increased risk for early death depends on form of disease and age of child during onset of disease. There is no cure for these disorders which currently draws upon experience from the field of childhood neuro disability.[3]

Ayurveda science gives detailed description about Vatavyadhi which can be merely co-related to nervous system disorders. Vata denotes for gati, chalata which carries affected other doshas and they are located to the site of Khavaigunya and produces disease. This is general pathogenesis of Vyadhi on the basis of ayurvedic science.

Case description

A six-year-old female child born first to second degree consanguineously married parents complaining of recurrent seizures, also unable to sit, unable to walk since 3-4 yrs. Associated complaints were unable to talk properly, detouring milestones, loss of appetite, dribbling of saliva. History of past illness was nothing significant. History of present illness stated that childwas apparently normal up to 2 years 9 month then child got seizure during sleep for which they approached to a local hospital. Seizure remains for longer time, child get admitted in hospital. Birth history of child was single live full term female child, delivered through LSCS, child cried immediately after birth, No history of NICU stay, Nothing significant after birth.



Developmental history was child attains all the milestones at appropriate age. Immunization history was appropriate to age only DPT booster of 5th year was not given. Family history states that her father had seizure at his age of 2-3 years which will get reduced afterwards.

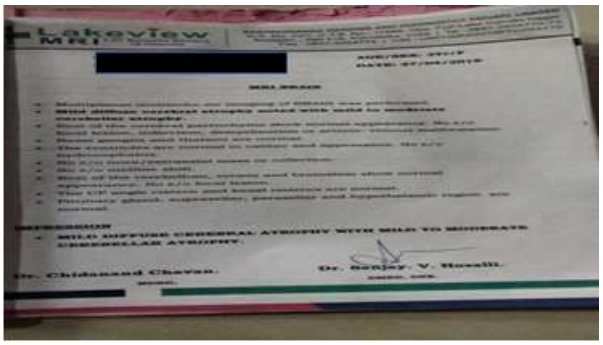
On examination child was having with seizure episodes, dribbling of saliva, loss of speech, unable to do motor activities. Child was totally bedridden and was on sodium valproate medication twice daily. From the investigation and examinations child was diagnosed as NCL-II.

General examination was within normal limits. Temperature- Afebrile(97.40F), Heart rate- 96/min, Respiratory rate 20/min. Systemic examination was child was conscious not oriented, Respiratory system mild ronchi were there, Gastrointestinal system on per abdominal examination was soft and non-tender, non-distension. Cardiovascular system was normal being S1/S2 sounds heard, no murmers.

Behalf of this ayurvedic Vyadhi pariksha denotes, Prakruti- Kapha-pitta, Vikruti-Vata, Sara- avara, Sahanana- Madhyama, Satmya-Sarvarasa, Satva-Heena, Vaya- Bala, Pramana- Madhyama, Vyayamashakti-Heena, Aharashakti- Madyama.

Asthvidha pariksha likewise Nadi- Pitta-kaphaja (82/min), Mutra-Prakrut, Mala-,Jivha-Alipta, Shabda-Aspashta, Sparsha-Anushnasheet, Druk(drushti)-Prakrut, Akruti- Prakrut.

On these basis Ayurvedic Vyadhi vinichaya can be explained. In this case the involved doshas are Vatapradhana tridosha, Dushya- Rasa, Majja and utrotar other dhatus, Sancharastana- Sarvasharir, Udbhavasthan-Shira, Vyaktastana- Sarvasharir Lakshana-Sarvagatravikshepa, Lalastrava, Vakastambha, Gatistambha Vyavachedaka Vyadhi- Apasmara, Sarvangvata Vyadhi vinischaya- Shiromarmabhighataja Vatavyadhi.



CONCLUSION

Batten disease is a rare genetic inherited disorder which starts at 5-6 years of life. The NCLs are a group of rare fatal neurodegenerative diseases. Most of the NCLs don't have approved disease-modifying therapies. There are currently several therapies also combination of multiple therapeutic approaches may be necessary to provide expected benefit. The therapeutic horizon for the NCLs is expanding. Multiple approaches have shown somewhat positive findings in pre-clinical studies. Presently, there is equipoise regarding the best methods of modifying these diseases. Because the NCLs represent multiple distinct pathobiological processes, it is likely that any single therapeutic approach will not be definitive for all forms. So, it is important that research continues across the spectrum from small to large-molecule and then gene therapy and beyond of that.

Shira is one among Trimarma, thus any minor injury to this organ may lead death or some dizardous symptoms. Shira was explained as Sadyopranahara marma. Acharya Sushrut explained that injury to shir may lead to death of the patient.[4]

Acharya Charak explained lakshanas of Shiromarmabhighata like neck rigidity, abnormal eye Movements, loss of movement, cough, closed eyelids, excess salivation, yawning, headache, less or total absence of speech. These symptoms can be co-related to this present case of Battens disease.

Chikistasutra of Shiromarmabhighataja vatavyadhi along with samanya vatavyadhi chikistasutra can be adopted for the present case. Chikista for shiromarmabhighata like Abhyang, Swedana, Upanaha, Snehapana, Basti, Avapidana nasya, Dhoompana with Vatahara dravyas.[5]

Clinical significance

NCL forms differ by the causative gene, gene product. Some of the affected proteins are soluble enzymes and others are membrane-bound proteins. There are no blood biomarkers that have been shown to correlate with disease progression in any form of. Potential approaches to disease modification include not also small-molecule drugs, large-molecule drugs, genetic medicines but also combination of therapies.

At the present time, the gene therapy is considered best potential to correct the underlying problem. Ultimately, based on complementary mechanisms of action, combination therapy may provide the best chance for meaningful disease modification. There is currently one disease-modifying ERT that has been approved by the US Food and Drug Administration (FDA) and the European Medicines Agency (EMA) for treatment of CLN2 disease.

Enzyme replacement therapy

ERT is a treatment module for deficiencies of enzyme that introduces purified recombinant enzymes through intracerebroventricular or intrathecal, intravenous injection. The injected enzyme is then delivered to the correct cellular compartment via receptor-mediated uptake. Deficiencies in soluble lysosomal enzymes leads to four of the Batten disease subtypes: CLN1 (PPT1), CLN2 (TPP1), CLN10 (CTSD) and CLN13 (cathepsin F (CTSF)).[4]

Gene therapy- For the treatment of neurodegenerative diseases and Lysosomal storage disorders, Adeno associated virus (AAV) – mediated gene therapy is promising option. It is effective in several models of Batten disease.[5]

Samanya vatvavyadhi Chikistasutra explained by Acharya Charaka is Snehana, Swedana, Mamsarasa, Upanaha, Snana, Seka, and other Shodhana modalities.[6] Shodhana modalities includes Virechana, Basti, Nasya etc.[7] These procedures may be useful for such conditions.

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