Dysplastic Myeloid Cells in a Case of Leigh Syndrome

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Abstract

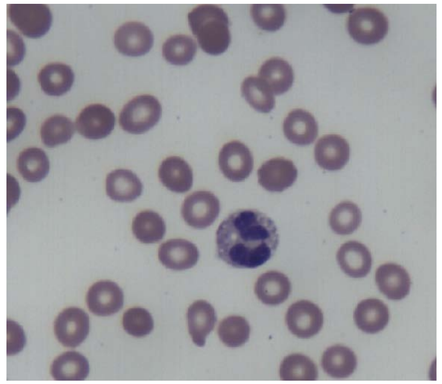
Subacute necrotizing encephalomyelopathy, also known as Leigh syndrome, is a rare disorder of infancy/childhood associated with altered mitochondrial metabolism. We describe a case of Leigh syndrome with unusual presence of dysplastic leukocytes in peripheral blood smear. Possible association between this patient's metabolic disorder and the dysplastic features seen in these leukocytes is suggested.

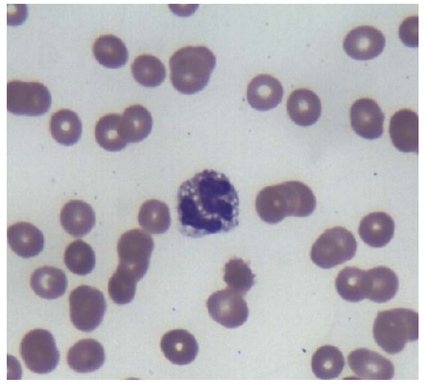
History/Microscopic Findings

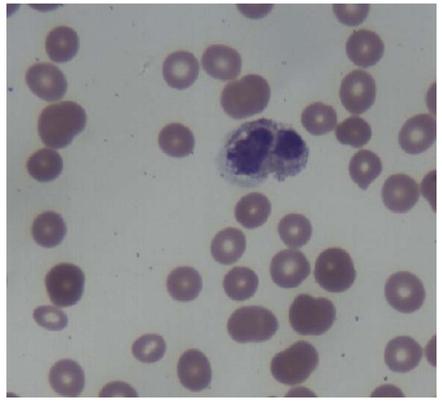
An eleven-year-old male patient with a known diagnosis of Leigh syndrome presented to the emergency department with an acute febrile illness, disseminated intravascular coagulation and possible sepsis. His complete blood count was significant for an elevated white blood count (14.4 x 10^9/µL), and decreased hemoglobin and hematocrit (10.4 g/dL and 30.8%, respectively). The patient's coagulation test results were also abnormal with elevated PT, PTT and FSP. The peripheral blood smear revealed numerous dysplastic cells of myeloid lineage (see figures, Wright stain, 100x).

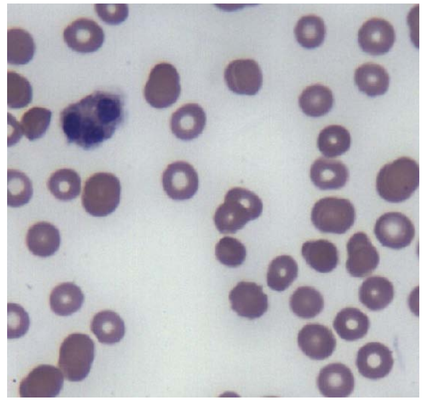
The dysplastic cells have pseudo-Pelger Hüet morphology, two or three nuclear segments that are connected by very thin nuclear strands. They also have numerous vacuoles and prominent granules. Furthermore, condensation of nuclear chromatin is observed.

Typical etiologies of dysplasia in the myeloid cells were considered and subsequently ruled out (myelodysplastic syndrome, medication, and chemical or toxic exposure)









Discussion

In Leigh syndrome, the biochemical defect in the respiratory chain most often involves respiratory complex IV (cytochrome c oxidase). The manifestations of this biochemical disturbance are primarily neurologic in nature. The disease is progressive and fatal, usually before the onset of the second decade of life. The current scientific and medical literature reveals the lack of a definitive link between Leigh syndrome and any specific hematologic abnormalities except for cytopenia.

The myeloid dysplasia in this patient most likely represents an unusual Leigh syndrome association that, to the best of our knowledge, has not been previously reported. Therefore, we suggest a possible association between this patient's metabolic disorder and the dysplastic features seen in the leukocytes.

References

-Ref 1

-Ref 2, etc.