

**48th ANNUAL DIAGNOSTIC SLIDE SESSION, 2007
DIAGNOSES AND REFERENCES**

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Case 2007-2

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Diagnosis: Primary (genetic) hemophagocytic lymphohistiocytosis (HLH), with a double heterozygote mutation of the perforin gene

Comment: During the child's hospital evaluation, serum C reactive protein (CRP) and C3 were elevated. Genetic studies were performed on tissue from brain and liver at autopsy, with the discovery of two missense mutations in the perforin gene. Secondary HLH is more common than genetic or familial, and the secondary form can be associated with Chédiak-Higashi syndrome, Griscelli syndrome and X-linked lymphoproliferative syndrome (XLP). Both genetic and secondary forms of HLH are triggered by an infection, often EB virus.

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