II ENCONTRO ASBAI – BRAGID

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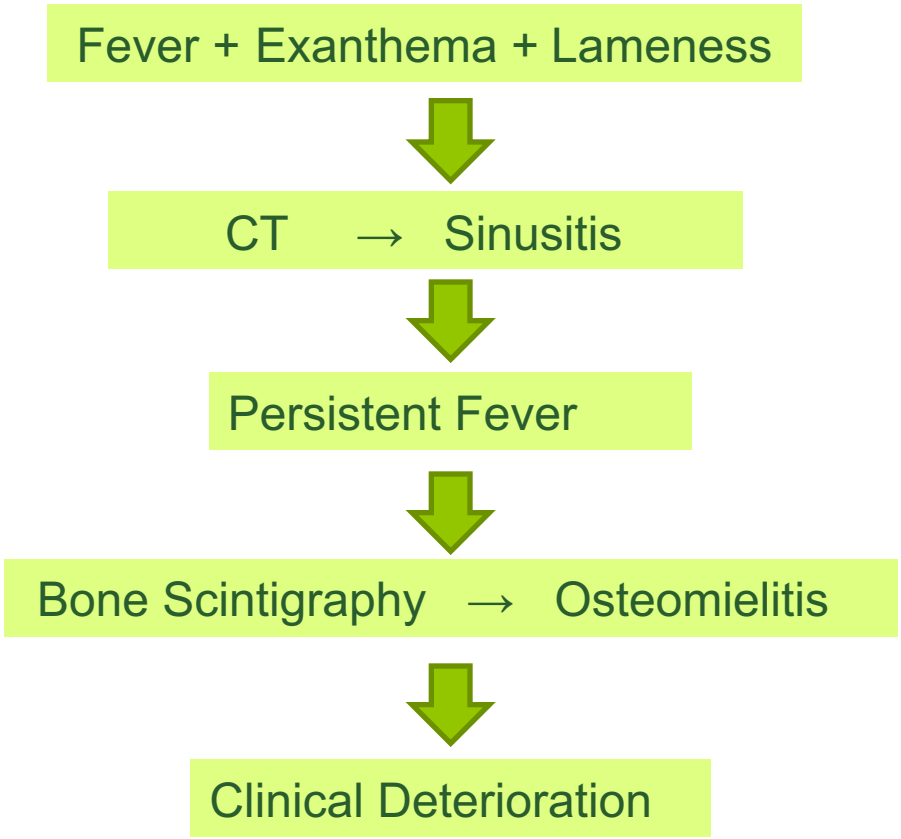
- FMS, male, born full-term
- Third child of non-consanguineous parents
- Symptoms started at 3 years old



Laboratory Findings at Diagnosis

Ferritin	6578 ng/ml
Triglycerides	368 mg/dl
Fibrinogen	200 mg/dl
Hemoglobin	8,9 g/dl
Leukocyte	5000 / m3
Platelets	53.000 / m3
DHL	1927 mg/dl
TGO/ TGP	1682 / 854 U/L

Abdomen CT = Hepatomegaly.

Myelogram – hypocellular bone marrow, hemophagocytosis

Diagnosis

Hemophagocytic Syndrome

- Immunologic, Rheumatologic, Oncologic, Infectious diseases was excluded
- Pulsotherapy was done for 3 days (Methylprednisolone), 1 month after the firsts symptoms (2015/jun)

- Genetic Panel: No pathogenic variants

ADA, IL2RG, IL7R, RAG1, RAG2, JAK3, CD3D, CD3E, CD3G, CD247, CD8A, NFKBIA, CIITA, DCLRE1C, FOXN1, LIG4, NHEJ1, PTPRC, PNP, RFX5, RFXANK, RFXAP, [illegible] CD19, IGLL1, LRRC8A, STAT3, MS4A1, AICDA, CD40, CD40LG, UNG, SH2D1A, PRF1, STX11, STXBP2, UNC13D, XIAP, [illegible]AS, FASLG, CASP8, CASP10, NRAS, FOXP3, IFNGR1, IFNGR2, IL12RB1, STAT1, ELANE, GFI1, HAX1, MPO, G6PC3, RAC2, CYBA, CYBB, NCF1, NCF2, NCF4, MYD88, SERPING1, WAS, WIPF[illegible], LYST, RAB27A, [illegible]ATA2, ORAI1, PIK3CD, MAGT1, STAT5B, EDA.

- He maintained pruriginous exanthema

3 days IVIG was done (2015/jun) with remission

(Ferritin = 43)

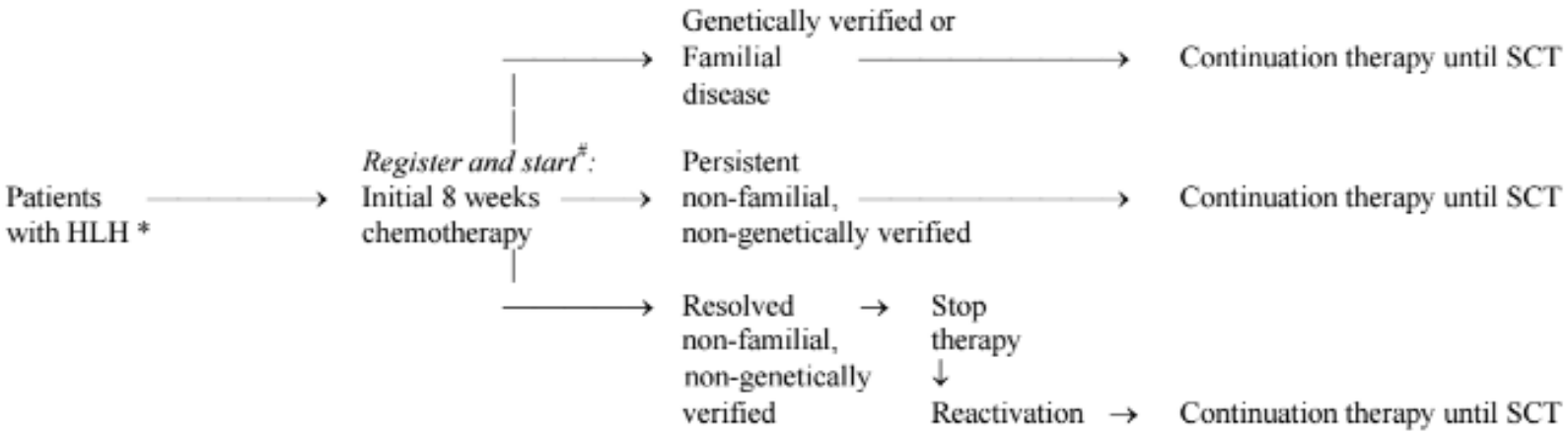
- 2015/ sept: Reactivation

Fever + Exanthema +

Increase Ferritin (4x basal) and trigliceryde

HLH 2004 Protocol was started

Figure 1: Flow-sheet for Children with Hemophagocytic Lymphohistiocytosis (HLH) in HLH-2004



* If there is a treatable infection it should be treated but be aware that this may not be sufficient and the patient may need HLH-treatment in addition. All severe forms should start HLH-treatment. If HLH is persistent or recurring consider that the patient may have an undiagnosed inherited disease. HLH may also develop secondary to a number of other diseases as malignancies, rheumatic diseases and metabolic disorders, requiring a different treatment.

Start therapy if the patient has a genetically verified disease, a familial form of HLH, or if the disease is severe, persistent, or recurrent.

HSCT was indicated

Avaliation pré- HSCT was done → CNS disease was diagnosed

MTX intratecal was started

HSCT was done in march,03 2016