KAWASAKI DISEASE WITH HEMOPHAGOCYTOTIC LYMPHOHISTIOCYTOSIS IN A CHILD WITH STXBP2 MONOALLELIC MUTATION

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INTRODUCTION

- ✓ **Secondary Hemophagocytic Lymphohistiocytosis (HLH)** is a rare and potentially life-threatening complication of Kawasaki disease (KD).
- ✓ This entity resembles primary HLH, a group of immune dysregulation syndromes, inherited in an **autosomal recessive** fashion.
- ✓ Genetic polymorphisms may play a central role in secondary HLH.

CASE REPORT

4-year-old white boy, no past medical history

D1 6-day history of **fever**, diffuse **erythema**, **odynophagia** and **right foot pain**.

Physical examination - Cheilitis. Strawberry tongue (Figure 1). Conjunctival hyperaemia. Perineal rash with desquamation. Feet oedema.

Initial blood work

- Haemoglobin 9.4g/dL; WBC 9600/uL; Platelets 103.000/uL
- ESR 35mm/h; Albumin 3,2g/dL; triglycerides 162mg/dL
- TGO: 19 U/L; TGP: 17 U/L; CRP 2.9mg/dL

Figure 1 – Strawberry tongue

Kawasaki Disease

Normal echocardiogram

Intravenous immunoglobulin (IVIG) and acetylsalicylic acid.

D3 Persistent fever → **2^a dosis IVIG** → Clinical Improvement
Apyrexia since D4

D7 Right ankle swelling

Surgical bone drainage
Cefuroxime iv (20 days)

MS Staphylococcus aureus

Clindamycin (10 weeks)

Figure 2 – MRI in D8 revealing distal tibial osteomyelitis

D20 Fever, exanthema and splenomegaly.

Hg 7,8g/dL; WBC 2320/uL (neut 80/uL)
Platelets 135000/uL
ALT 221 U/L; ferritin 36036ng/mL;
Fibrinogen 247mg/dL; sCD25 3346U/ml.

Hemophagocytic syndrome

- Reference unit.
- Bone marrow biopsy negative for hemophagocytosis

Conservative approach was assumed → Gradual clinical and analytical normalization

9 months after discharge

Bone marrow examinations - **Hemophagocytic cells**

- no other hemophagocytic criteria
- **Regular outpatient vigilance**

Monoallelic STXBP2 mutation

DISCUSSION

- ✓ Type 5 Familial HLH is caused by biallelic STXBP2 mutation. **Heterozygosity may grant hemophagocytosis susceptibility**, when triggered by conditions, such as KD or infections.
- ✓ Optimal approach guidelines and specific HLH criteria for KD are necessary.
- ✓ We highlight the importance of **genetic study in these patients**, in order to anticipate possible future recurrence.

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