EFFICACY AND SAFETY OF INFLIXIMAB IN NF-KB ESSENTIAL MODULATOR DELETED EXON 5 AUTO-INFLAMMATORY SYNDROME: A CASE REPORT

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Abstract number: 5PSQ-108

ATC code: L04-IMMUNOSUPPRESSANTS

Background

The NF-KB essential modulator deleted exon 5 auto-inflammatory syndrome (NEMO-NDAS) is an X-linked auto-inflammatory disease belonging to the systemic auto-inflammatory diseases (SAIDs). NEMO-NDAS affects the skin (ectodermal dysplasia) and the immune system. A few cases have been reported in France.

Aim and objectives:

To Describe the use and safety of infliximab in NEMO-NDAS in a 9-month-old child

Case description

We report a **9-month-old baby** who initially presented a **long-lasting fever** and a **panniculitis**. No infectious nor autoimmune causes were found, and the interferon signature was low. A **corticosteroid treatment** was started.
Further genetic analyses showed an anomaly of the NEMO gene compatible with a **NEMO-NDAS**. Several pathways are modified in this disease, including the interferon pathway.
No recommendations nor relevant literature for specific treatment was found.
As it the NF-KB is known to be regulated by the tumor necrosis factor (TNF), an anti-TNF agent has been introduced : **infliximab**.

Results

Figure 1: Schematic representation of the link between autoinflammatory and autoimmunity diseases.
Marcuzzi A & all. Autoinflammatory Diseases and Cytokine Storms-Imbalances of Innate and Adaptative Immunity. Int J Mol Sci. 2021 Oct 18;22(20):11241. doi: 10.3390/ijms222011241. PMID: 34681901; PMCID: PMC8541037.

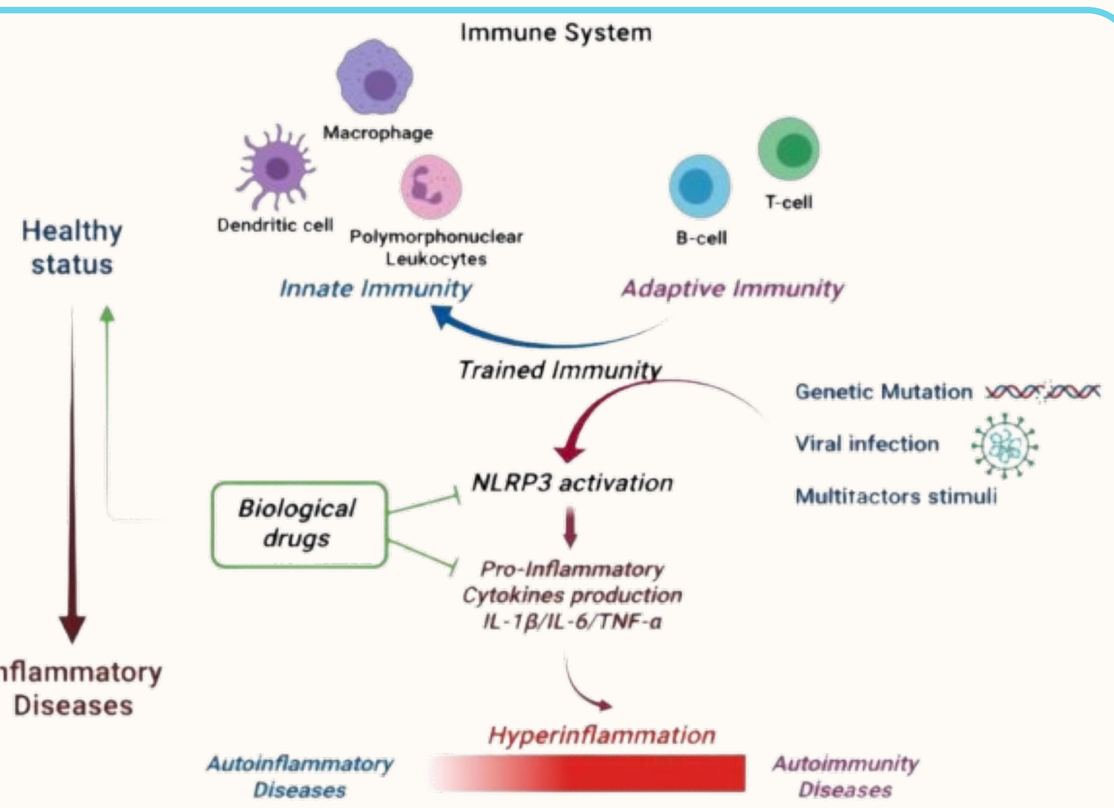

Figure 1

Pre-therapeutic assessment:
- QuantiFERON-tuberculosis: negative
- Up-to-date vaccinations
- Serologies HBV, HIV and EBV: negative

Week 0: introduction of infliximab at a dose of 5 mg/kg:

Prednisone dosage = 7.5 mg	Clinically: Indurated, circumscribed erythematous skin lesions, painful on palpation	Biological inflammatory syndrome: - C-reactive protein (CRP) = 93 mg/L - Sedimentation rate (SR) = 23 mm - Serum amyloid A (SAA) protein = 504 mg/L	Well-tolerated cure

Week 2: infliximab 5 mg/kg:

Prednisone dosage = 2 mg	Clinically: No cutaneous manifestation, no intercurrent events	Biological inflammatory syndrome: - CRP = 1 mg/L - SR = 2 mm	Well-tolerated cure

Week 4, 8 & 12: infliximab 5 mg/kg:

Prednisone stopped	Clinically: No cutaneous manifestation but some lymphadenopathy, no intercurrent events	Biological inflammatory syndrome: - CRP = 1 mg/L - SR = 2 mm - SAA protein < 6.9 mg/L	Well-tolerated cure

Week 16: infliximab 5 mg/kg:

Prednisone stopped	Clinically: No cutaneous manifestation but inflammatory foot oedema, no intercurrent events	Biological inflammatory syndrome: - CRP = 1 mg/L - SR = 2 mm - SAA protein < 6.9 mg/L	**Impossible to perform an infusion** (catheter placing impossible)

END of infliximab, switch to adalimumab due to the availability of its subcutaneous form

Conclusion and relevance

Infliximab was used **successfully** in our case and led to **remission** in 2 weeks with **good tolerance and no adverse effect**. Infliximab seems to be a well-tolerated treatment option for **NEMO-NDAS in infants**.
The contribution of the **clinical pharmacist** could promote therapeutic education in patients with rare diseases.