

CYTOTOXIC T LYMPHOCYTE ANTIGEN 4 MUTATIONS ON T REG AND ABATACEPT: A PAEDIATRIC CASE REPORT

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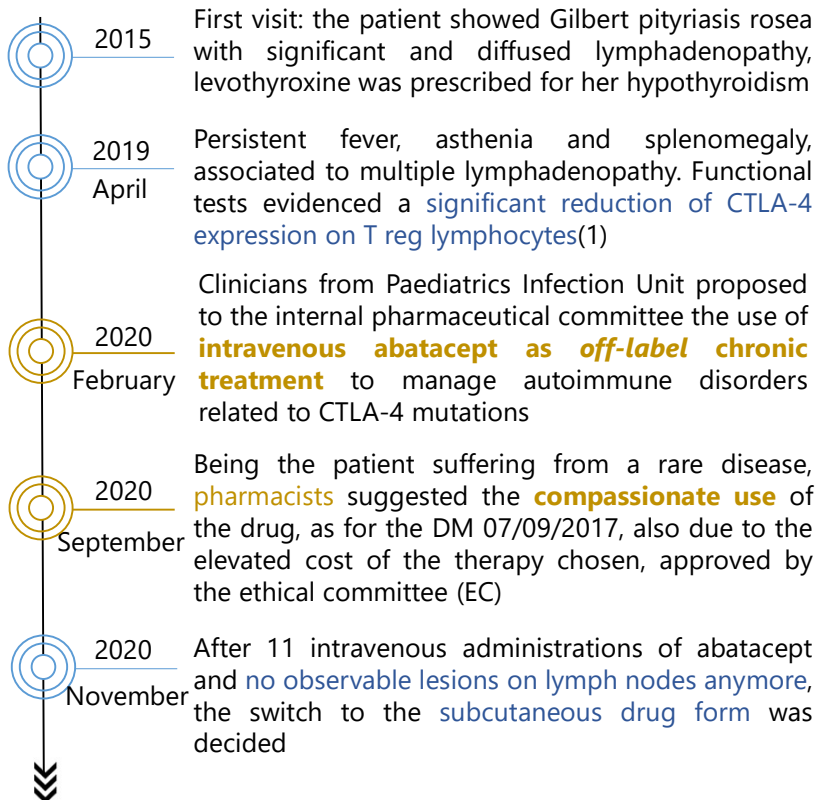


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Background and importance

The lymphoproliferative autoimmune syndrome caused by **cytotoxic T-lymphocyte antigen 4 (CTLA-4) insufficiency** is characterized by immune dysregulation, haploinsufficiency and multi-organ disorders. This condition is classified as rare disease and **no drugs** are registered for the treatment yet. → **Abatacept**, an anti-rheumatic agent, selectively modifies and inhibits a key-stimulating signal which can activate T-lymphocytes. The effect produced is a dose-dependant reduction on serological interleukins (2 and 6), TNF α and other factors preventing T-lymphocytes activation.

Material and methods



Aim and objectives

To report the clinical record of a 15-year-old female patient presenting a VOUS mutation on CTLA-4, causing multiple lymphadenopathies, Gilbert pityriasis rosea, hypothyroidism, chronic urticaria, angio-oedema and fever, asthenia and splenomegaly lastly.

Results

The use of abatacept shows positive outcomes to date (after twenty-two doses), observing **regression in all lymphadenopathy sites**. Moreover, the EC approval of the **subcutaneous** form makes the therapy chosen **self-manageable and administrable at home**, surely simplifying the girl's routine.

Conclusion and Relevance

Abatacept use in CTLA-4 mutations on T reg may represent a valid chance with positive disease regression. The observation of a single patient does require more studies and applications to detect a possible systematic use of the drug for the treatment of this rare condition. Further studies will be implemented to study and analyse the long-term effect of abatacept on VOUS mutations of T reg.

References

(1) Schwab et al. J Allergy Clin Immunol. 2018;142(6):1932-46.

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Immunosuppressive agents