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), TNFα and other factors preventing T-lymphocytes activation.

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 drug 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