Dear all,

With this message, I would like to ask you for your collaboration regarding the establishment of a CTLA4 cohort.

We are currently in the process of identifying as many patients with CTLA-4 deficiency as possible to get a better understanding of the clinical phenotype of those patients and to determine the best treatment options for them. To date, sirolimus and abatacept, as well as HSCT have been applied, but the outcome is variable.

So far our cohort consists of 38 CTLA4 mutation carriers, of which 27 are symptomatic, as well as 11 asymptomatic carriers. The latest age of onset was at age 49. Apart from the already published phenotype including hypogammaglobulinemia, enteropathy and lymphadenopathy we have observed lymphoma and HLH in several of our patients. This illustrates the importance of gathering more data on this enigmatic disease.

Please find our publication on CTLA-4 deficiency attached, the website for this disorder is: www.ctla4-deficiency.org.

If you take care of any patients with the genetic diagnosis of CTLA-4 deficiency or if you have patients with a phenotype that might indicate CTLA4 mutations as the underlying genetic defect, please contact b.grimbacher@ucl.ac.uk, or bodo.grimbacher@uniklinik-freiburg.de, and we will send you more information on how to participate in this study.

The participation in this collaborative effort shall of course lead to a collaborative publication, but shall not prevent you from publishing your case/cases in smaller case reports/case series separately.

Best regards,

Bodo

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