Background and Importance Epidermolytic ichthyosis (EI) is a skin genetic disorder that predominantly affects joints and friction areas with a limited number of therapeutic options (topical and systemic treatments). In some severe EI subtypes, the off-label use of ustekinumab is based on marked elevation of cytokines in the Th17/IL-23 pathway, similar to inflammatory diseases like psoriasis.

Aim and Objectives To evaluate the efficacy and security of ustekinumab in an 8-year-old female patient with severe EI with KRT1 mutation, refractory to topical treatment and oral retinoids.

Material and Methods Firstly, she underwent keratolytics and emollients with scarce clinical response. Due to the absence of effectiveness of these topical therapies, she received oral retinoids (acitretin) up to maximum tolerated dose, but some erythematous lesions and hyperkeratosis rapidly appeared on her skin. For this reason, the treatment had to be discontinued several times. In April 2022, Dermatology Service requested the off-label use of ustekinumab with a dosage of 0.75 mg/kg at weeks 0, 4, 8 and 12, and then administered each 12 weeks to the Pharmacy therapeutics committee.

Afterwards, an extensive review was carried out by Pharmacy Service. A report was made with a positive assessment for approval of treatment. This decision was supported by some case reports showing clinical results of ustekinumab in some EI subtypes and the lack of available alternative therapies in this case.

Results At the beginning of treatment with ustekinumab, extensive scaly erythematous lesions with circinate margins were observed, affecting the facial area, trunk and extremities, accompanied by diffuse palmoplantar keratoderma.

After three months of the first administration of ustekinumab, the patient was examined by a dermatologist. An excellent clinical response was observed with resolution of the facial lesions and almost complete on the trunk, with hyperkeratotic lesions persisting in folds, without underlying erythema. Moreover, no adverse events related to ustekinumab were registered.

Conclusion and Relevance Ustekinumab is suggested to be an alternative therapy in some severe EI subtypes refractory to topical and systemic treatments. In spite of being safe and effective in this patient, longer studies are needed to consider ustekinumab in the therapeutic management of EI.

REFERENCES AND/OR ACKNOWLEDGEMENTS
Conflict of Interest No conflict of interest.