THE DARK SIDE OF APDS2: A STORY TOLD THROUGH FDG-PET

Catelli A[1], Nanni C[2], Mulè R[3], Pession A[4], Conti F[4]

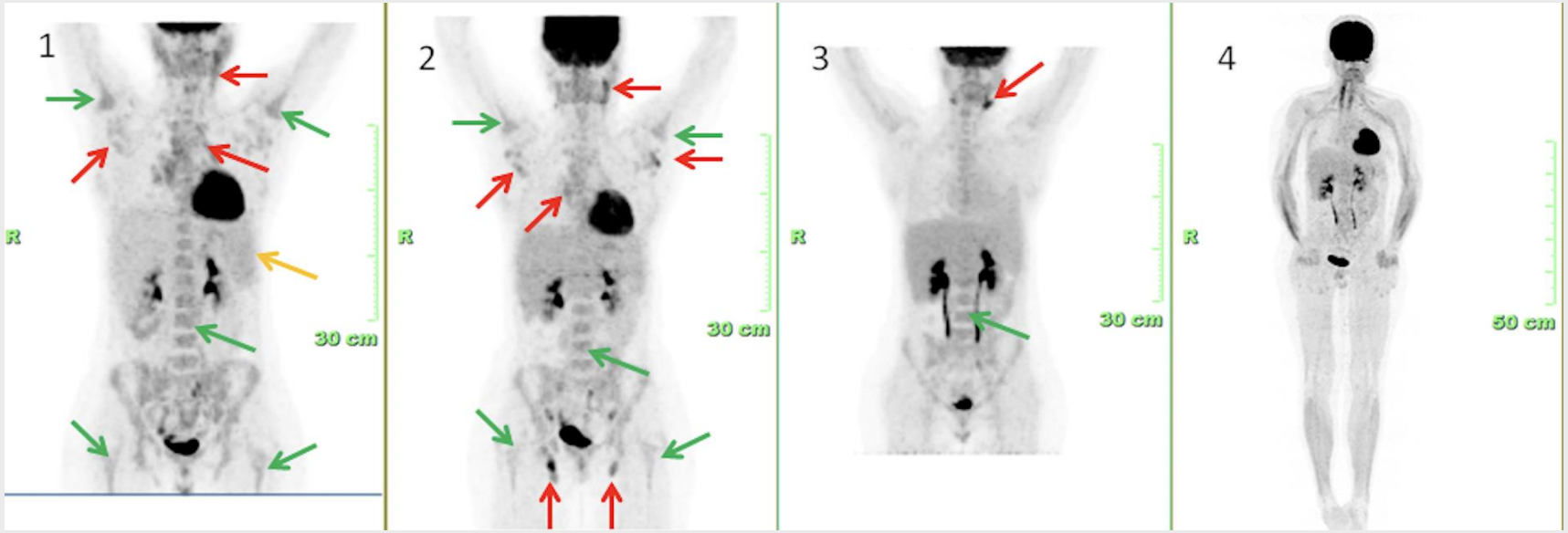
[1] Specialty School of Paediatrics, Alma Mater Studiorum-University of Bologna, Bologna, Italy, [2] Nuclear Medicine, IRCCS Azienda Ospedaliero-Universitaria di Bologna, Bologna, Italy, [3] Rheumatology Unit, Azienda Ospedaliero-Universitaria di Bologna, Bologna, Italy, [4] Pediatric Unit, IRCCS Azienda Ospedaliero-Universitaria di Bologna, Bologna, Italy

Introduction

Lymphoproliferation and increased risk of malignancy are key features of activated phosphoinositide 3-kinase-δ syndrome 2 (APDS2)[1]. We report the use of FDG-PET/CT as a helpful diagnostic tool for accurate identification of inborn errors of immunity (IEI), especially among lymphoproliferative disorders[2].

Case report

Activating mutation of *PIK3R1* was found in a 30-year-old woman with refractory systemic lupus erythematosus, autoimmune cytopenia, and diffuse lymphoproliferation. Ten years before, our patient was diagnosed with Hodgkin's lymphoma, although atypical aspects emerged: FDG-PET/CT at baseline showed several hot supra and subdiaphragmatic lymph nodes with a very symmetrical distribution, associated to inhomogeneous increased tracer uptake in axial and appendicular bone marrow and slight and diffuse increase of the spleen metabolism *(Figure 1).* Several scans repeated after chemotherapy and autologous stem cell transplantation revealed incomplete resolution, with persistent diffuse increased FDG uptake at multiple supradiaphragmatic nodes and axial and appendicular bone marrow *(Figure 2-3).* One year after APDS2 diagnosis and rapamycin treatment, FDG-PET/CT confirmed remission of lymphoproliferation, showing a complete metabolic normalization of all sites *(Figure 4).*



Figures: MIP (Maximum Intensity Projection) Image of FDG PET/CT at staging (1), after transplantation (2-3) and after rapamyicin treatment (4). Green arrows: hot bone marrow. Red arrows: hot lymphnodes. Yellow arrow: abnormal spleen metabolism.

Discussion

FDG-PET/CT has a crucial role in differentiating malignant proliferation from immune dysregulation phenotypes. In our case, inadequate response to lymphoma treatment was due to diffuse lymphoproliferation, and numerous imaging studies could be avoided.

Therefore, the presence of atypical patterns and unusual metabolic uptake at FDG-PET/TC should be interpreted as a red flag for the need of an early immunological evaluation, especially in patients with other features of immune dysregulation.

References:
[1] Jamee M, et al. Clinical, Immunological, and Genetic Features in Patients with Activated PI3Kδ Syndrome (APDS): a Systematic Review. *Clin Rev Allergy Immunol.* 2020;59(3):323-333.
[2] Zanoni L, Mattana F, Calabrò D, et al. Overview and recent advances in PET/CT imaging in lymphoma and multiple myeloma. Eur J Radiol. 2021;141:109793.