Erdheim-Chester disease with concomitant Rosai-Dorfman like lesions: a distinct entity mainly driven by MAP2K1

Jérôme Razanamahery,1,2 Eli L. Diamond,3 Fleur Cohen-Aubart,1,2* Karl-Heinz Plate,4 Giota Lourida,5 Frederic Charlotte,6 Zofia Hélias-Rodziewicz,7,8 Gaurav Goyal,9 Ronald S. Go,9 Ahmet Dogan,10 Omar Abdel-Wahab,10 Benjamin Durham,10 Neval Ozkaya,10 Zahir Amoura,1,2 Jean-François Emile7,8 and Julien Haroche1,2*

JR, ELD and FCA contributed equally to this work

1Internal Medicine Department 2, Assistance Publique-Hôpitaux de Paris, French National Reference Center for Histiocytoses, Pitié-Salpêtrière Hospital, Paris, France; 2Paris VI University, UPMC, Sorbonne Universités, Paris, France; 3Department of Neurology, Memorial Sloan Kettering Cancer Center, New York, NY, USA; 4Institute of Neurology, Goethe University Hospital, Frankfurt Cancer Institute, German Consortium for Translational Cancer Research, Frankfurt, Germany; 5Department of Internal Medicine and Infectious Disease, Sotiria Hospital, Athens, Greece; 6Department of Pathology, Assistance Publique-Hôpitaux de Paris, Pitié-Salpêtrière Hospital, Paris, France; 7Pathology Service, Ambroise Paré Hospital, Assistance Publique-Hôpitaux de Paris Boulogne, Paris, France; 8EA4340, Université de Versailles SQY, Université Paris Saclay, Boulogne, Paris, France; 9Division of Hematology, Mayo Clinic, Rochester, MN, USA and 10Department of Pathology, Memorial Sloan Kettering Cancer Center, New York, NY, USA

Correspondence: JULIEN HAROCHE - julien.haroche@aphp.fr.

doi:10.3324/haematol.2019.246937
Supplementary data:

Histology was performed on 4 micrometer thick tissue sections after setting with Hematoxylin & Eosin and immunohistochemistry, which included at least CD1a, S100 and [CD68 or CD163] primary antibodies. All slides were reviewed by pathologists experienced in histiocytosis.

Detection of mutations was performed on tissue biopsies infiltrated by histiocytosis. Tumor DNA was extracted from formalin-fixed and paraffin embedded tissues. Detection of $BRAF V600E$ mutation was performed as described in Diamond et al. or Melloul et al. Detection of mutations of other genes was performed by targeted next generation sequencing.

Samples in France were analyzed using MiSeq (Illumina) after preparing the libraries with the Custom Amlicon Low Input Kit. The targeting genes were: $AKT1$, $ALK$, $ARAF$, $ASXL1$, $BRAF$, $CALR$, $CBL$, $CDK4$, $CDKN1B$, $CDKN2A$, $CEBPA$, $CSF3R$, $CTNNB1$, $DNMT3A$, $EGFR$, $EZH2$, $FLT3$, $GATA2$, $GNA11$, $GNAQ$, $GNAS$, $HERC1$, $HRAS$, $IDH1$, $IDH2$, $JAK2$, $JAK3$, $KIT$, $KRAS$, $KTM2D$, $MAML3$, $MAML3D1$, $MAP2K1$, $MAP2K2$, $MAP2K3$, $MAP2K4$, $MAP2K6$, $MAP3K1$, $MAP3K8$, $MAP3K9$, $MAP3K10$, $MAP3K19$, $MAP4K4$, $MAPK1$, $MAPK11$, $MAPK9$, $MPL$, $NF1$, $NOTCH1$, $NOTCH2$, $NPM1$, $NRAS$, $PDGFRA$, $PIK3CA$, $PP6C$, $PTEN$, $PTPN11$, $RAC1$, $RAF1$, $RIT1$, $RUNX1$, $SETBP1$, $SRSF2$, $STAG2$, $STK19$, $SYNGAP1$, $TAOK1$, $TAOK2$, $TET2$, $TP53$, $U2AF1$, $WT1$, $ZRSR2$.

Samples in MSKCC were analyzed with IMPACT or whole exome sequencing.

Search of mutation has not been performed on peripheral blood nor bone marrow biopsy.

References

Diamond et al. Cancer Discov 2016

Melloul et al. Haematologica 2018

Diamond et al. Nature 2019