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

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Supplementary data:

Histology was performed on 4 micrometer thick tissue sections after setinaing 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, MAMLD1, 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