Molecular understanding of histiocytic sarcoma

21 Archival cases of primary histiocytic sarcoma

- Whole exome sequencing
- RNA-sequencing

21/21 A high number of genetic alterations within the RAS-RAF-MAPK pathway

6/21 Alterations in NF1
5/21 Alterations in MAP2K1
3/21 Concurrent mutations
4/21 Alterations in PTPN11
4/21 Alterations in BRAF
4/21 Alterations in KRAS
1/21 Alterations in NRAS
1/21 Alterations in LZTR1

5/7 Cases with alterations in NF1 and/or PTPN11 had disease involving the gastrointestinal tract

Egan et al., Haematologica, 2020