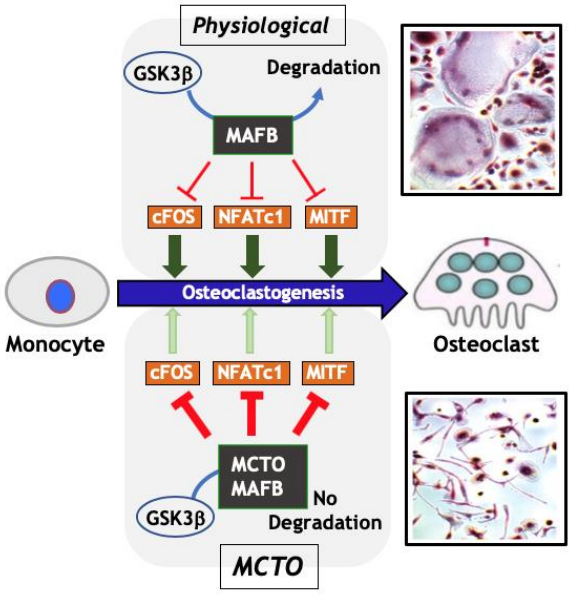
Studies of the **Myeloid Cell Biology** Group led by Dr. Ángel Corbí have focused on **Multicentric Carpotarsal Osteolysis syndrome** (MCTO), a very rare genetic disorder that belongs to the group of "vanishing bone" skeletal dysplasias, characterized by the progressive loss of bones in hands and feet (usually the carpal bones of the wrist and tarsal bones of the ankle). Besides, MCTO is associated with kidney disease in more than 50% of patients, and progresses rapidly to end-stage renal failure. Also genetic, this pathology shows an autosomal dominant inheritance pattern with heterozygous mutations in the MAFB gene, which codes for a transcription factor of the "large MAF" family (MAFB). In addition, the MCTO resembles the recently defined **Aymé-Gripp syndrome**, a very rare disease characterized by autism spectrum disorder, sensorineural hearing loss, skeletal involvement and cardiac abnormalities, which is also caused by heterozygous mutations in the MAF-related MAF gene.

Studies developed by the group on the functional specialization of human macrophages have shown that MAFB and MAF are specifically expressed in human anti-inflammatory macrophages and that MAFB is essential for the acquisition of the anti-inflammatory profile of human macrophages.[1] In fact, MCTO macrophages were found to be impaired in their ability to generate functional osteoclasts (bone-degrading macrophages).[2]

To facilitate the development of "macrophage reprogramming" therapeutic strategies for chronic inflammatory diseases, this laboratory is currently working on the identification of genes and functions specifically controlled by MAFB in human macrophages, as well as in the search for other naturally-occurring mutations in MAFB and MAF genes as a mean to fully understand the pathophysiological role of both factors during immune and inflammatory responses.



Regulation of expression and function of MAFB under physiological conditions and hypothetical deregulation in the case of MCTO (from *Cuevas et al., J Immunol, 198:2070-2081, 2017*).

[1] Sierra-Filardi et al., The Journal of Immunology, 2014, 192: 3858–3867
[2] Cuevas et al., The Journal of Immunology, 2017, 198:2070-2081