
1. **Shedding light on complex clinical cases with the SOPHiA Whole Exome Solution.** Pantelis Constantoulakis

2. **Diagnostic microdeletion/microduplication detection by exome sequencing enabling copy number variation analysis.** G. Christopoulou[1], A. Oikonomaki[1], S. Samara[1], L. Florentin[2], F. Sachinidi[2], S. Vittas[3], P. Constantoulakis[1]

3. **Next Generation Sequencing (NGS) as a key player in improving diagnostic yield for rare diseases (RDs): the collaborative experience of 2 centres in Greece.** N. Marinakis, G. Christopoulou, D. Veltra, M. Sviggou, K. Kekou, C. Sofokleous, E. Tsoutsou, K. Kosma, A. Oikonomaki, P. Constantoulakis, H. Fryssira, J. Traeger-Synodinos