Phenotype-genotype correlation in pediatric HCM : a CMR study

Elena Panaioli 1,2 , Volodia Dangouloff-Ros 2 , Nathalie Boddaert 2 , Damien Bonnet 1 , Diala Khraiche 1

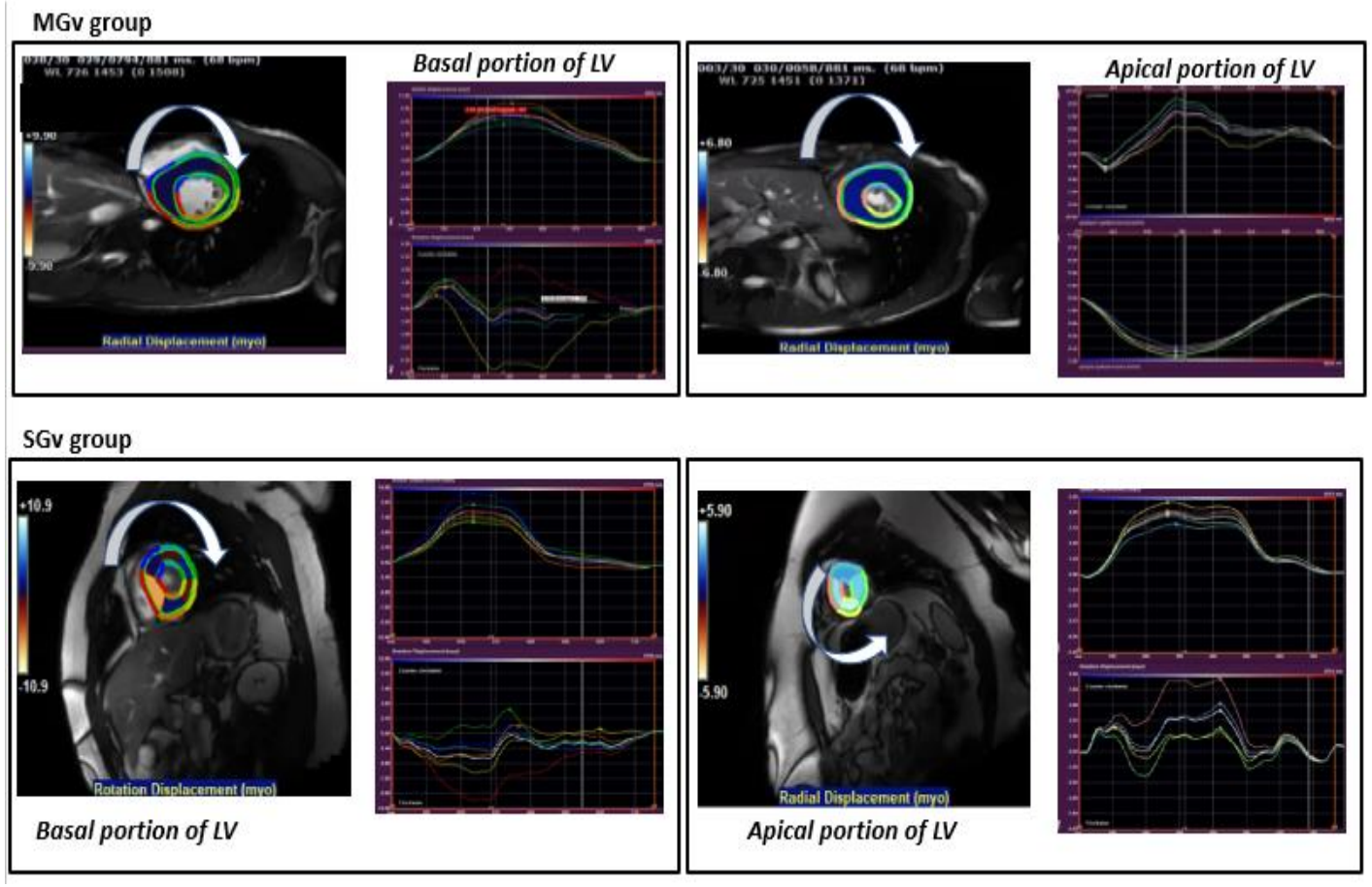
1 . Unité médico-chirurgicale de Cardiologie Congénitale et Pédiatrique, Centre de Référence Malformations Cardiaques Congénitales Complexes - M3C, Hôpital universitaire Necker-Enfants malades, France , **2.** Paediatric Radiology Department, Hôpital Necker Enfants Malades, Paris, France; UMR 1163, Imagine Institute, Paris, France

Purpose: The aim of our study is to assess differences of LV and LA mechanics features, in pHCM, between patients harbouring multiple pathogenic or likely pathogenic variants (MGv, n=16) or single genetic variations (SGv, n=41), by FT-CMR.

Methods: 57 patients (age at CMR 11,6+/- 4,1 years).

FT-CMR : LA, LV feature tracking (FT) derived strain and LV twist (LVT) .

LV twist was calculated as the difference between apical and basal rotation.



Conclusions: Patients with multiple genetic variants have a greater LV mass and altered LV mechanics with reduced LV twist . This study gives insights in phenotype-genotype correlation in paediatric HCM and warrants larger longitudinal studies to assess its clinical significance

Results: In MGv group, the indexed LV mass was greater (108.8 +/-53.0) than in SGv group (74.3+/- 22.2) (p=0.03). In MGv group a LGE pattern was found in 75 % and in SGv group in 54% patients . LV FT derived strain values were not statistically significant different between the groups (MGv vs SGv: GLS -15.8+/-5.3 vs -18.7+/-4.8, GCS -27.8+/8 vs -31.1+/-8.6, GRS 44.7+/-24.6 vs 62.3+/-32).

LVT was reduced in MGv group (0.04+/-7.6) vs (7.4+/-7.4) in SGv (p=0.003).

LA contractile function did not differ between the groups.