

**NF1 myocardial mass and cardiac function echocardiography measurements are similar to unaffected controls.**

**Presenting author: Juliana Souza, MD**

Institution/University: *Neurofibromatosis Outpatient Reference Center, School of Medicine, Federal University of Minas Gerais, Belo Horizonte, Brazil*

**Background** - Aerobic capacity ( $VO_{2max}$ ) reflects the functionality of respiratory, muscular and cardiovascular systems [Lee D-c et al, 2010]. It has been observed that individuals with NF1 have normal pulmonary function, respiratory muscle weakness [Souza et al, 2012], reduced maximal handgrip force (Fmax) [Souza et al, 2009], earlier vascular involvement [Friedman et al, 2002] and decreased  $VO_{2max}$  [Souza et al, 2013]. To better understand the cardiac component of  $VO_{2max}$  in individuals with NF1, we performed echocardiography measurements of their myocardial mass and cardiac function.

**Aim** - To compare NF1 myocardial mass and cardiac function (measured by Doppler echocardiography) with unaffected controls matched by sex, age and physical activity levels.

**Methods** – Data was obtained by transthoracic echocardiography of 20 individuals with NF1 (11 male, 9 female) and 21 unaffected controls (10 male, 11 female). Systolic and diastolic cardiac function was evaluated using conventional M Mode, 2D and Doppler echocardiographic indices (ejection fraction – EF and left ventricular fractional shortening – LVFS). EF was calculated using the Teichholz method and left ventricle myocardial mass (LVMM) obtained by American Society of Echocardiography-Penn convention formula (Vivid 7 GE scanner, Horten, Norway®). Strain measurements were performed offline, with dedicated automated software EchoPac PC, version 11.1.0. GE®. Each LV wall was divided in 3 segments and a global systolic longitudinal strain (GSLs) was obtained. Data was presented as mean and standard deviation and variables were compared using Student *t* test. For multiple comparisons, analysis of variance was used. Probability values < 0.05 were considered statistically significant.

**Results**- There was no statistically significant difference in LVMM (indexed to body surface area) in controls ( $75.5 \pm 13.2$  g/m<sup>2</sup>) compared to individuals with NF1 ( $74.1 \pm 18.7$ ) ( $P=0.793$ ). Radial cardiac function measured as EF (%) and LVFS (%) was statistically greater in NF1 group ( $70.7 \pm 5.8$ ;  $39.7 \pm 4.3$ ) than in control group ( $67.5 \pm 3.5$ ;  $36.8 \pm 2.6$ ) ( $P=0.025$ ;  $0.008$ ), after adjustments for weight (without clinical implications that could explain the lower  $VO_{2max}$  previously observed in individuals with NF1). No difference in GSLs was observed between the two groups ( $-20.3 \pm 2.05$ , NF1;  $19.8 \pm 1.4$ , controls) ( $P= 0.427$ ).

**Conclusion** – Individuals with NF1 presented normal cardiac mass as well as myocardial radial and longitudinal function when compared with unaffected controls.

**Full list authors:** Souza JF, MD; Sette JBC, MD; Pena JLB, Ph.D.; Rezende NA, Ph.D.; Rodrigues LOC, Ph.D.

**Granting agencies:** FAPEMIG, CAPES and CNPq.

**References:**

1. Lee D-c et al. Mortality trends in the general population: the importance of cardiorespiratory fitness. *J Psychopharmacol.* 2010; 24(11): 27-35.
2. Souza JF et al. Pulmonary function in individuals with NF1: a preliminary report. In: NF Conference 2012: Proceedings of the Children's Tumor Foundation Conference; Jun 8-12, New Orleans, LA, USA. 2012; p. 71.
3. Souza JF et al. Muscular force is reduced in neurofibromatosis type 1. *J Musculoskelet Neuronal Interact* 2009; 9(1):15-7.
4. Friedman JM et al. Cardiovascular disease in neurofibromatosis 1: a report of the NF1 Cardiovascular Task Force. *Genet Med.* 2002; 4(3): 105-1.
5. Souza JF et al. Exercise capacity impairment in individuals with neurofibromatosis type 1. *AJMG.* 2013; 161A (2):393-5.