

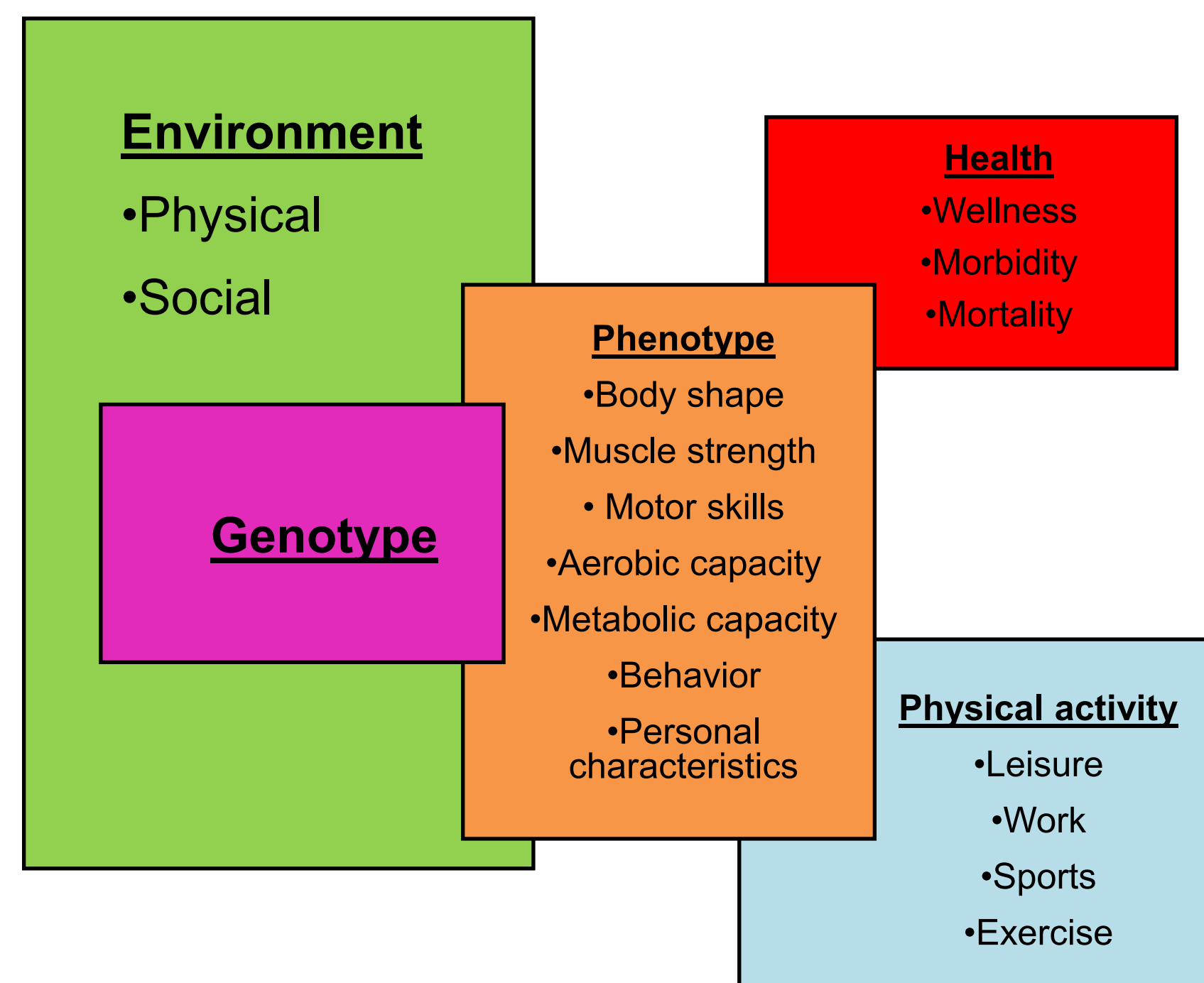


# Aerobic capacity is reduced in Neurofibromatosis type 1: a preliminary report

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**Background :** Neurofibromatosis type 1 (NF1) presents a wide variety of clinical features, including muscular hypotony. Reduced maximal voluntary handgrip muscular force ( $P=0.001$ ) has been observed among patients attended at the Neurofibromatosis Outpatient Reference Center of Minas Gerais Federal University (CRNF), Brazil (Souza et al, 2009). It is known that aerobic capacity (maximal oxygen uptake/ $VO_{2max}$ ) is determined by cardiovascular capacity and general muscle metabolism and strength (Bouchard & Shepard, 1994). Furthermore,  $VO_{2max}$  is directly related to life expectancy and quality of life (Blair et al, 1995). Patients with NF1 have not only a shorter lifetime but also a lower quality-of-life (Wolkenstein et al, 2001). However, to our knowledge,  $VO_{2max}$  has not been quantified among NF1 patients.

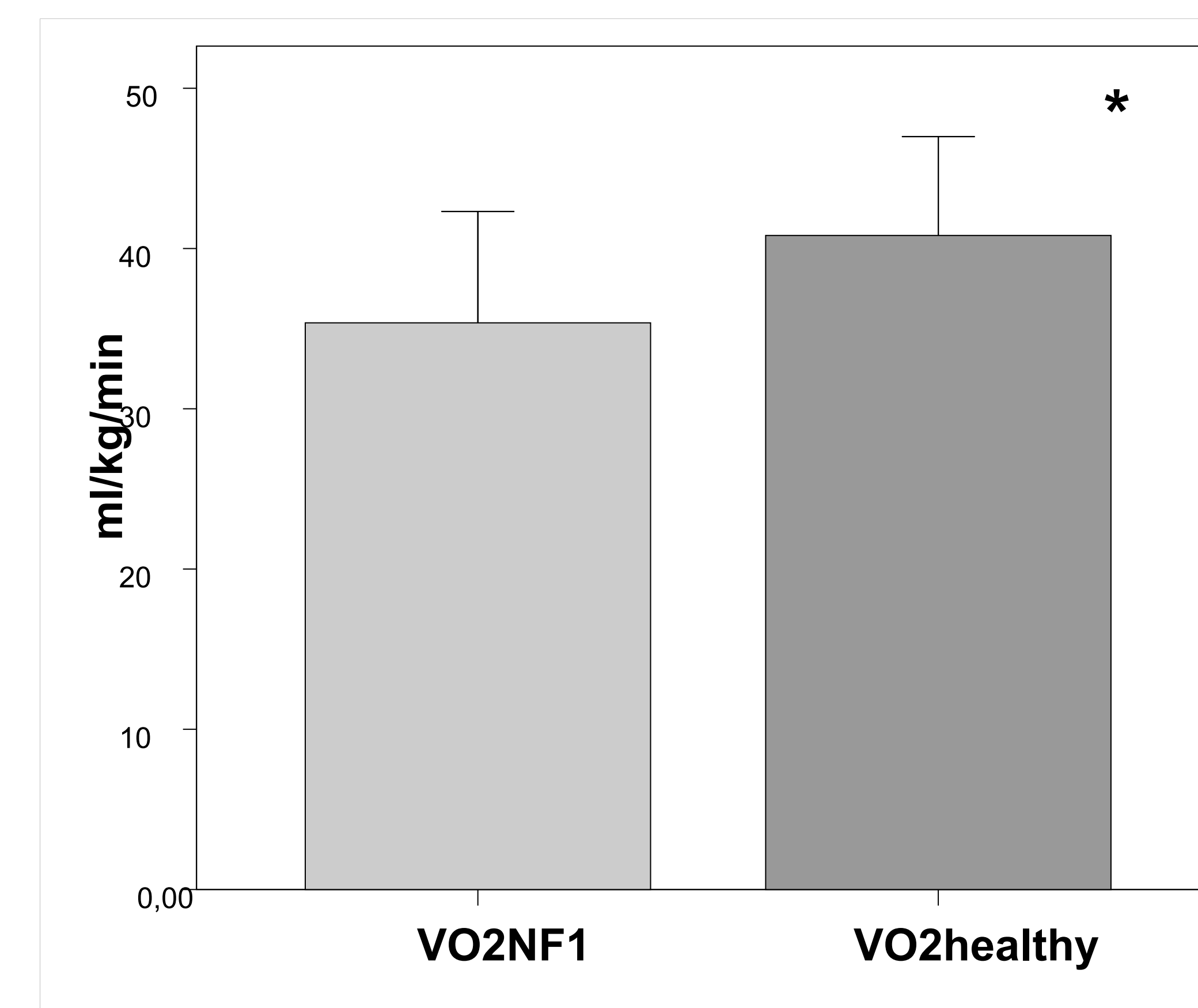


**Figure 1. Relationship between physical activity, fitness and health.** The genotype is expressed in a particular phenotype depending on the physical and social environment. The phenotype is characterized by specific behaviors (including the pattern of physical activity), which feed back the expression of the genotype on a more fitted phenotype. Phenotype's adaptability to the environment influences health, which interferes with the willingness for physical activity and, consequently, the expression of the phenotype itself. Modified Shepard & Bouchard (1994)

**Aim:** To compare maximal oxygen uptake ( $VO_{2max}$ ) of NF1 patients with age and sex matched healthy volunteers (HV).

**Methods -** Seventeen volunteered NF1 patients (NIH criteria, 5 male, 12 female), aged between 18 and 58 years, without heart and/or lung involvement or physical limitations to the test, were matched by sex and age to 17 HV. All volunteers signed the informed consent protocol.  $VO_{2max}$  was estimated from a maximal exercise capacity treadmill test using Bruce's protocol. Data were compared using a Student *t* Test.

**Results-** The mean  $VO_{2max}$  in HV ( $40.8 \pm 6.2$  ml/kg min) was greater than in NF1 patients ( $35.5 \pm 6.5$  ml/kg min) ( $P=0.02$ ) (Figure 1). As expected, healthy male showed greater  $VO_{2max}$  ( $46.0 \pm 5.2$  ml/kg min) than healthy female ( $38.2 \pm 5$  ml/kg min) ( $P=0.02$ ) and this gender difference was just a tendency among male and female NF1 patients ( $P=0.047$ ).



**Figure 2. Aerobic capacity** expressed as maximal oxygen uptake ( $VO_{2max}$  in ml/kg/min; showed as mean  $\pm$  SD) in NF1 patients ( $n=17$ ) and in healthy individuals ( $n=17$ ) matched by sex and age. (\*):  $P<0.02$  for healthy  $VO_{2max} >$  NF1  $VO_{2max}$

**Conclusion:** Aerobic capacity is reduced in NF1 patients compared to healthy subjects matched for sex and age.

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