

Genes and altitude I

High altitude populations

Chronic mountain sickness: **EPO, EPO-receptor, HIF-1a, von Hippel-Lindau**, prolyl hydroxylase domain containing 1, 2, 3, and phosphatase and tensin homolog deleted on chromosome 10: *no association* (Mejia, 2005)

Aldosterone (gene CYP11B2) : -344T/C, intron 2 conversion Iw/Ic, and A5160C *over expressed* in Himalayan natives (Rajput, 2006)

HIF1 α (intron 13) – GT15 *over expressed* in Japanese/GT14 *over expressed* in Sherpas) (Suzuki, 2003)

HIF 1 α , VEGF, eNOS, iNOS (UVEC) : *no difference* of expression between Tibetans and Hans (Gao, 2005)

Genes and altitude II

Animal species adapted or not to high altitude

- **cytochrome c oxidase** subunit II (shift of D by T at 115 position) in camelids (Di Rocco, 2006)
- Similarities of **HIF 1 α** in the Yak and other species (Dolt, 2007)
- Up or down-regulation of **eIF-2 α , eIF-4, notch gene homolog-1, MAD homolog-4, calpain** in the heart of mice exposed to chronic or intermittent hypoxia (Fan, 2005)
- **chromosome 2 (GGA2), GGA4 and GGA6** and pulmonary hypertension in the chicken (Rabie, 2005)

Genes and altitude III

Subjects with AMS or HAPE or pulmonary hypertension (PH)

- **Renin-angiotensin** system (A(1166)C and G(1517)T single-nucleotide polymorphisms in AT(1)R gene) *associated to HAPE in Japanese, but not ACE-I/D polymorphism (Hotta, 2004)*
- **ACE and Angio II receptor**(ACE (ACE(A-240T), dbSNP rs4291; and ACE(A2350G), dbSNP rs4343), the intronic Alu insertion in ACE (ACE I/D), and the SNP ATR(A1166C), (dbSNP rs17231380) in AGTR1d) and AMS in Nepalese : *no association* (Koehle, 2006)
- **ACE, tyrosine hydroxylase, serotonin transporter [5-HTT], endothelial NO synthase [eNOS]** genes : *no association to HAPE* (Mortimer, 2004)

Genes and altitude IV

Subjects with AMS or HAPE or PH

- **ACE** polymorphisms I/I, D/D, I/D : *no association* with AMS (Dehnert, 2003), polymorphism I/I more frequent in Kirghyz residents with PH (Aldashev, 2002), polymorphism D/D less frequent in COPD patients with RV hypertrophy (van Suylen, 1999)
- **Serotonin transporter** (genotype LL) *associated with PH* in COPD (Eddahibi, 2003)
- **eNOS**: (Glu298Asp variant and 27-base pair (bp) variable numbers of tandem repeats) *associates to HAPE* (Droma, 2003), wild alleles of the Glu298Asp and eNOS4b/a polymorphisms *more frequent* in Sherpas than in Nepalese (Droma, 2006)
- **Proteins A1 and A2 of the surfactant** (SP-A1 (C1101T, T3192C, and T3234C) and SP-A2 (A3265C)) *associated to susceptibility to HAPE* (Saxena, 2005)
- **hsp70-2 and hsp-hom** (+190 G/C, +1267 A/G, 2437 G/C) : *association* between hsp70-2 B/B, hsp70-hom A/B and B/B and AMS/HAPE in Chinese (Zhou, 2005)

Genes and altitude V

Athletes trained at altitude or sea level

- **ACE and EPO** in endurance athletes at moderate altitude : *no association* (Gonzalez, 2006)
- **ACE/DD** *associated* to LV hypertrophy in endurance athletes (Hernandez, 2003)
- **EPO** (marker D7S477), 3434 C --> T polymorphism in the 3' HRE sequence, and variability of erythropoietic response to altitude : *no association* (Jedlickova, 2003)