OSTEOPOROSIS

PATHOGENESIS	
	The bone mineral density (BMD) is the major risk of fracture in people with osteoporosis and it represents the interaction of genetic, metabolic and environmental factors. Epidemiological studies demonstrated how the osteoporosis has a strong genetic component (the hereditary of the BMD is about the 50-80%). The genetic research concentrated both on the research of responsible gene of rare monogenic bone diseases, and on the identification of polymorphisms on involved gene in the bone metabolisms, which are associated to an increased risk of osteoporosis.
POLYMORPHISM COL1A1 SP1 AND THERAPY WITH BISPHOSPHONATE.	
	Genes COLIA1 and COLIA2 codify for the main protein of the bone matrix: the collagen of type I. Associations among polymorphism Sp1, BMD and osteoporosis fracture are present in many populations. Recent studies demonstrated how the genotype COLIA1 takes to a difference in BMD, creating a 1.5-3 times increased risk of fractures. The same polymorphism (COLIA1 Sp1) was associated to others relevant phenotypes for osteoporosis, such as the loss of bone mass after the menopause, the geometry of the femoral neck and the answer to therapy with di-phosphonates. The answer to therapy with di-phosphonated, which must be calculated measuring the BDM of the femoral neck, is very different between genetic groups SS (wild-type) and Ss or ss (mutated). In SS group there's an increase of the BMD of the 1.32% after 3 years of treatment, while in Ss or ss group there's a decrease of the 0.66%.
ESTROGEN RECEPTOR (ER) POLYMORPHISM.	
	The polymorphism Pvull (PP) on ER was associated to a decreased BMD. This polymorphism is directly correlated to a low number of TA repetitions on the promoter of the gene ER. In a study on Italian women it was possible to correlate it to the BDM. (a low number of repetitions of the dinucleotide TA is associated to the low BMD and to an high fracture risk). The presence of the polymorphism Pvull increases the fracture risk about three times.
VITAMIN D RECEPTOR (VDR) POLYMORPHISM.	
	One of the identified polymorphism on VDR (BsmI, alleles B/b) was associated to the osteoporosis. The allele BB was indeed found with a three times greater frequency in patients with osteoporosis than in a checked population. The BMD results to be under the average in 74% of homozygotes patients for the allele B (BB), but superior to the average in 64% of homozygotes patients for the allele b (bb). In patients with a polymorphism PP (Pvull-/-), the presence of the polymorphism BB (Bsml -/-) on VDR is linked to a decreased BMD.

Laboratorio di diagnostica molecolare

Further information or bibliographic references can be asked to the laboratory.

OSTEOPOROSIS

TEST	
	Highlight of polymorphisms by means of PCR and restriction analyses.
SAMPLE TAKING	
	Blood/EDTA, 2 ml.
EXECUTION	
	Daily.
COST	
	Upon request.

Further information or bibliographic references can be asked to the laboratory.