Context The adenomatous polyposis coli (APC) tumour - suppressor protein controls the Wnt signalling pathway by forming a complex with glycogen synthase kinase 3beta (GSK - 3beta), axin / conductin and betacatenin.	Facts (Sampled) (complex, subclass of, protein) (GSK, instance of, protein) (glycogen, instance of, protein) (APC, instance of, protein)
Context Recently, we reported five Austrian families with generalized atrophic benign epidermolysis bullosa who share the same COL17A1 mutation.	Facts (Sampled) (epidermolysis bullosa, instance of, mutations)
Context We identified four germline mutations in three breast cancer families and in one breast - ovarian cancer family. among these were one frameshift mutation, one nonsense mutation, one novel splice site mutation, and one missense mutation.	Facts (Sampled) (frameshift mutation, subclass of, Germline mutations) (Nonsense mutation, subclass of, Germline mutations) (splice site mutation, subclass of, Germline mutations) (missense mutations, subclass of, Germline mutations) (Nonsense mutation, subclass of, cancers) (frameshift mutation, subclass of, cancers) (missense mutations, subclass of, cancers)
Context A nonsense mutation in exon 17 (codon 556) of the RB1 gene was found to be present homozygously in both the retinal and the pineal tumours.	Facts (Sampled) (retinal, instance of, gene) (Nonsense mutation, subclass of, gene)
Context Sixteen different p16 germline mutations were found in 21 families, while one germline mutation, Arg24His, was detected in the CDK4 gene.	Facts (Sampled) (p16, subclass of, Germline mutations) (Germline mutations, subclass of, gene) (p16, instance of, gene)
Context Aspartylglucosaminuria (AGU) is a rare disorder of glycoprotein metabolism caused by the deficiency of the lysosomal enzyme aspartylglucosaminidase (AGA).	Facts (Sampled) (Aspartylglucosaminuria, subclass of, deficiency)
Context Detection of heterozygous carriers of the ataxia - telangiectasia (ATM) gene by G2 phase chromosomal radiosensitivity of peripheral blood lymphocytes.	Facts (Sampled) (ATM, instance of, gene) (G2 phase, part of, blood) (G2 phase, instance of, gene)
Context HLA typing for HLA - B27, HLA - B60, and HLA - DR1 was performed by polymerase chain reaction with sequence - specific primers, and zygosity was assessed using microsatellite markers.	Facts (Sampled) (microsatellite, subclass of, primers) (DR1, instance of, microsatellite) (microsatellite, subclass of, typing)