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Epigenetics in Sporadic Retinoblastoma

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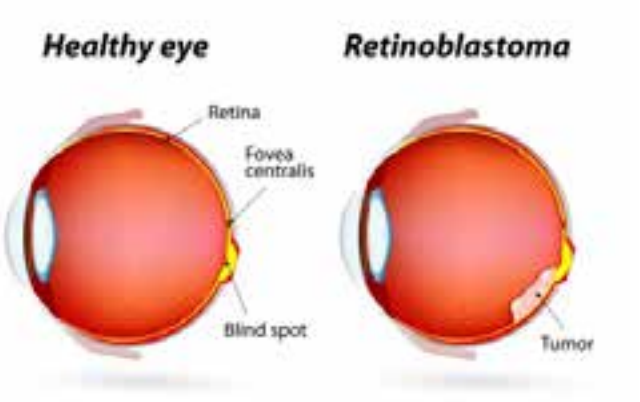
Retinoblastoma (Rb) is a relatively rare cancer, although represents the most common ocular malignancy in children. Tumor rapidly develops from the immature cells of the retina and it can be aggressive and potentially fatal. Rb occurs in one (unilateral) or both eyes (bilateral) and can spread beyond the affected orbit, presenting leukocoria, strabismus, proptosis and other atypical signs. It is classified as hereditary or sporadic (non-hereditary) forms. Due to its high invasiveness, the clinical practice for Rb often results in enucleating the affected eye, with severe loss of vision. Rb presents unique psychosocial and financial challenges to patients, especially in developing countries where it shows higher incidence and lower survival rate. Rb was the first tumor to draw attention to the genetic etiology of cancer and its initiation and progression, traditionally seen as a genetic disease, are currently involved in epigenetic abnormalities along with genetic alterations. Some epigenetic modifications play an important role in Rb tumorigenesis: gene silencing could represent a somatic event and it is then observed in about 10% of unilateral sporadic Rb. Biallelic aberrant methylation of CpG islands of the promoter region of tumor-suppressor gene RB1/p105 and other Rb family members (genes p107 and RB2/p130) can result in complete loss of function and induce uncontrolled cellular proliferation. Abnormal hypermethylation of additional tumor-suppressor and pro-apoptotic genes has been characterized in Rb, demonstrating an epigenetic component to gene inactivation. Methylation of transposable elements adjacent to the genes with metastable epialleles and methylation of regulatory elements of imprinted genes are other epigenetic modifications occurring during retinoblastoma development, involved in silencing gene expression. Moreover, some cancer-related genes exhibited correlative histone modifications in Rb, when compared to normal tissue. In conclusion, sporadic retinoblastoma is the most commonly accepted form of the non-hereditary disease, suggesting that is an epigenetic, rather than a genetic disease.



Healthy eye (left) vs Intraocular Rb (right)(Ref. National Cancer Institute)

Recent Publications

1. Pileri P et al. (2016) FAT1: a potential target for monoclonal antibody therapy in colon cancer. Br J Cancer 115(1):40-51.
2. Naldi I et al. (2014) Novel epigenetic target therapy for prostate cancer: a preclinical study. PlosOne 9(5): e98101
3. Taranta M et al. (2011) Magnetically driven bioreactors as new tools in drug delivery. J BABM S5:1-8.
4. Cinti C et al. (2011) Newly engineered magnetic erythrocytes for sustained and targeted delivery of anti-cancer therapeutic compounds. PlosOne 6(2): e17132.

Biography

Ilaria Naldi has completed her Master's degree in Biological Sciences (Physio-pathological field) after graduating from the University of Florence, Italy. She has her main expertise in preclinical research, with 10-years of working experience as Research Scientist in the therapeutic areas of experimental oncology, epigenetics, and drug-delivery systems, bacterial and fungal infectious diseases. She collaborated as Visiting Scientist with the Marshall University, USA and then she obtained the Clinical Research Associate certification. She completed her PhD degree in Biomedical Sciences (Experimental and Clinical Oncology) and she has been registered as Italian Professional Biologist. She also recently attended the Master in Pharmaceutical Management and Marketing, while working as Clinical Research Coordinator for Phase II, III Clinical Trials in Onco-Hematology (Lymphoma Division) at AOU Careggi in Florence.

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