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Chemical mutagenesis: Generation of novel mutants and elucidation of functional pathways in the eye

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With the exception of trauma and infectious disease, the majority of the reported eye diseases are genetic in nature. While some of these diseases have associated animal models, many do not. Through a chemical mutagenesis program, Translational Vision Research Models, we have identified 89 ocular models with defects ranging from cataracts to photoreceptor degeneration. The molecular basis has been elucidated in 54 mutants. Of these, 25 were remutations with some similarities to previously published alleles, 13 were remutations with at least one significant difference to previously reported models, 5 were caused by mutations in genes found in human patients but for which no mouse model had been described, and 12 were caused by mutations in novel genes that were previously not implicated in eye disease. The disrupted genes encode for proteins involved in retinal developmental, in cellular structure or adhesion, in cellular trafficking, in metabolism, in the visual cycle, in synaptic signaling, and in post-translational processing of retinal proteins. Chemical mutagenesis can be, not only, used to generate new disease models but can also be used in sensitized mutagenesis-driven modifier screens. In mice, this can be a powerful approach to reveal molecules/pathways that disrupted in the disease state, and as a consequence this approachcan potentially identify new treatment targets. A large number of disease-modified strains have been identified in a sensitized screen of B6/B6N-*crb1*^{rd8}/*crb1*^{rd8} mice. Enhanced retinal lesions, neovascularization, and pigmentation phenotypes have been documented in the strains established from the sensitized screens.

Biography

Patsy M Nishina, Professor, is currently working at The Jackson Laboratory. Her laboratory is actively involved in generating or identifying mouse models that carry mutations, which lead to retinal diseases. In the later case, models are acquired through screening either standard mouse strains for spontaneous mutations or ENU mutagenized mice. Classical genetic as well as molecular, biochemical, imaging, and immunological approaches are used to identify molecules and pathways that are important in retinal development, maintenance and function within these models. She is particularly interested in understanding how disruption of molecules and pathways affect RPE and Mueller glia cell development, function and maintenance.

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