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An analysis of type I collagen from the oim model mouse

An analysis of type I collagen from the oim model mouse Kristin Rolwes¹, Stephanie Carleton², and Charlotte Phillips³, PhD 1 LSUROP, 2 Genetics Area Program, 3 Departments of Biochemistry and Child Health, University of Missouri-Columbia. Project Abstract Osteogenesis imperfecta (OI) type III is a heritable disorder leading to impaired connective tissue function in type I collagen containing tissues leading to bone fragility, blue-grey sclera, short stature, and hearing loss (1). Normal type I collagen is a heterotrimeric molecule containing two proalpha1(I) collagen chains and a similar but genetically distinct proalpha2(I) collagen chain. The osteogenesis imperfecta murine (oim) model mouse produces only homotrimeric type I collagen due to a single nucleotide deletion in the COL1A2 gene resulting in a non-functional proalpha2(I) collagen chain. The oim mutation is bred on the B6C3Fe background strain of mice. The result is expression of an abnormal type I collagen molecule which leads to the above phenotype (1). The severity of phenotype varies greatly between affected individuals even between family members with the same mutation. This indicates that another factor besides the mutation in the collagen gene is impacting the phenotype. This impact is believed to be caused by modifier genes which affect the overall phenotype of those affected. This study is aimed at investigating the primary differences between two mouse models with differing backgrounds. Both the C57BL/6J and B6C3Fe background strains were selected for this experiment due to differences in their genetic makeup. The C57BL/6J strain is a congenic background strain while the B6C3Fe is a hybrid background strain. The C57BL/6J animals were bred to the B6C3Fe animals with the (OI) type III mutation leading to the presence of the oim mutation on the C57BL/6J background. Preliminary investigation hopes to show differences in the production of type I collagen between each strain. Once the initial differences in each strain are identified at the phenotypic level future studies include analyzing specific differences in gene expression which may be analyzed by microarray for the presence of possible affecting modifier genes. This portion of the project deals directly with the analysis of type I collagen in both oim and wildtype mice of the B63Fe background strain. 1. Chipman S.D. et al. Proc. Natl. Acad. Sci. USA 90:1701-05, 1993.