ATTITUDES AND PRACTICES OF GENETIC COUNSELORS IN PROVIDING PREDICTIVE TESTING TO MINORS AT RISK FOR LI-FRAUMENI SYNDROME

Publication No. ———— *

Allison Copeland, B.S.

Supervisory Professor: Louise Strong, MD

Li- Fraumeni Syndrome (LFS) is a rare autosomal dominant hereditary cancer syndrome caused by mutations in the *TP53* gene that predisposes individuals to a wide variety of cancers, including breast cancer, soft tissue sarcomas, osteosarcomas, brain tumors, and adrenocortical carcinomas. Individuals found to carry germline mutations in *TP53* have a 90% lifetime cancer risk, with a 20% chance to develop cancer under the age of 20. Despite the significant risk of childhood cancer, predictive testing for unaffected minors at risk for LFS historically has not been recommended, largely due to the lack of available and effective screening for the types of cancers involved. A recently developed screening protocol suggests an advantage to identifying and screening children at risk for LFS and we therefore hypothesized that this alongside with the availability of new screening modalities may substantiate a shift in recommendations for predictive genetic testing in minors at risk for LFS. We aimed to describe current screening recommendations that genetic counselors provide to this population as well as explore factors that may have influenced genetic counselors attitude and practice in regards to this issue. An online survey was emailed to members of the National Society of Genetic Counselors (NSGC) and the Canadian Association of Genetic Counsellors (CAGC). Of an estimated 1000 eligible participants, 172 completed surveys that were analyzed. Genetic counselors in this study were more likely to support predictive genetic testing for this population as the minor aged (p<0.05).
This trend was influenced by the individual requesting testing, the presence of a family history of LFS-related childhood onset cancers, and the availability of appropriate risk management (p<0.05). Counselors were knowledgeable regarding recent screening measures which may be helpful in directing physicians towards new options. When discussing their attitudes towards predictive testing, genetic counselors considered many factors and raised both the opportunity for benefit and harm, while also addressing the gap in knowledge on this subject. These findings indicate the relevance of continued discussion on the appropriateness of predictive genetic testing in this population and the importance of continued research and education to develop future guidelines.