FAMILY HISTORY OF A GENETIC CONDITION REFERRAL GUIDELINE

MAINE MEDICAL PARTNERS - PEDIATRIC SPECIALTY CARE (DIV. OF GENETICS) • 1577 CONGRESS ST, PORTLAND, ME • (207) 662-5522

HIGH RISK

SUGGESTED EMERGENT CONSULTATION

SYMPTOMS AND LABS

Neurodegenerative or progressive disorders with onset in infancy/ childhood

Treatable conditions (including inborn errors of metabolism)

First degree family member deceased of a genetic condition

SUGGESTED PREVISIT WORKUP

We will require documentation of DNA testing or how a specific diagnosis was established in an affected family member(s)

Please contact the Geneticist on call or one of the Genetic Counselors at (207) 662-5522 option 8 to discuss

Please provide as much information as is known or where additional information may be obtained

MODERATE RISK

SUGGESTED CONSULTATION OR CO-MANAGEMENT

SYMPTOMS AND LABS

Most referrals for concerns of a genetic condition in family history will fall into this category

Common examples include aortic root aneurysms, cardiomyopathy, NF, other known genetic disorder

Note that not all genetic conditions have available clinical testing and not all insurance companies will pay for genetic testing

SUGGESTED WORKUP

Please include documentation of pertinent exam findings, other consult reports

We will need documentation of DNA testing or how the diagnosis was made in an affected family member(s) in order to provide informative guidance

Provide as much detail as possible, as well as contact information for your office in case we need additional specific information

LOW RISK

SUGGESTED ROUTINE CARE

SYMPTOMS AND LABS

There are some commonly ordered genetic tests for which results have no clinical significance (e.g. MTHFR polymorphisms)

Some genetic diagnoses do not warrant referral of children < 18 years of age, (e.g. Hemochromatosis, carrier testing for ANY diagnosis, most adult onset diagnoses)

SUGGESTED MANAGEMENT

Testing of minors/children for Adult Onset Genetic Disorders or to determine Carrier status is not ethically appropriate, however the family may benefit from Genetic Counseling regarding risks

CLINICAL PEARLS

- **ALL referrals are reviewed by an ABGC Certified Genetic Counselor and/or a Geneticist; we will expedite any referral when medically indicated.**
- Early diagnosis and testing is only appropriate for children if the Genetic Condition that is in their family has medical implications in childhood (including screening, surveillance or treatment).
- If the patient is currently pregnant, contact MMP Women's Health Maternal Fetal Medicine at (207) 771-5549
- If the family history is of cancer, contact the MMC Cancer Risk and Prevention Program at (207) 396-7787

Maine Medical PARTNERS

2/17