POLE, POLD1, GREM1
HEREDITARY COLORECTAL CANCER

4 THINGS YOU NEED TO KNOW

1. Patients with a personal and/or family history of colorectal cancer should consider genetic testing that includes POLE, POLD1, and GREM1.

2. Patients with a germline mutation in POLE, POLD1, or GREM1 are at increased risk for multiple colorectal polyps and possibly other cancers.

3. Patients with a POLE, POLD1, or GREM1 mutation should consider increased screening for colorectal cancer such as earlier and more frequent colonoscopies.

4. Family members of patients with a mutation may be at an increased risk for cancer and should consider genetic testing.

WHICH PATIENTS SHOULD CONSIDER GENETIC TESTING FOR POLE, POLD1, AND GREM1?

Consider testing for these and other colorectal cancer related genes for patients with a:

- Personal history of polyposis (multiple colorectal polyps)
- Personal history of colorectal cancer under 50 years old
- Family history of a close relative with colorectal cancer under 50 years old

HOW DOES A POSITIVE RESULT IMPACT MY PATIENT’S CANCER RISK?

<table>
<thead>
<tr>
<th>Gene</th>
<th>Lifetime Colorectal Cancer Risk</th>
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<tbody>
<tr>
<td>POLE</td>
<td>Increased risk$^{1-5}$</td>
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<tr>
<td>POLD1</td>
<td>Increased risk$^{6,7}$</td>
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<tr>
<td>GREM1</td>
<td>Increased risk$^{8,9}$</td>
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WHAT ARE THE MEDICAL MANAGEMENT RECOMMENDATIONS FOR A PATIENT WITH A MUTATION IN POLE, POLD1, OR GREM1?

- Start colonoscopies at 25-30y and frequency depends on polyp burden
- Surgical evaluation if appropriate

*Adapted from national guidelines

WHAT ARE THE IMPLICATIONS FOR MY PATIENT’S FAMILY?

- First degree relatives (parents, siblings, and children) of a patient with a POLE, POLD1, or GREM1 mutation have a 50% chance to have the same mutation
- Additional relatives may also be at risk and should consider genetic testing under the advisement of a healthcare professional

TESTS THAT INCLUDE POLE, POLD1, AND GREM1:

<table>
<thead>
<tr>
<th>ColoNext</th>
<th>CancerNext</th>
<th>CancerNext-Expanded</th>
<th>CustomNext-Cancer</th>
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REFERENCES