

DEVELOPING NEW CLINICAL RECOMMENDATIONS FOR HEREDITARY POLYPOSIS IN CHILDREN

Practical Guidance for Management of Pediatric Patients

Polyps are relatively common in children and typically do not signal a risk of serious illness. However, in some children who have several polyps or a suspicious family history, an increased cancer risk is quite possible. Most pediatric gastroenterologists see few patients with a major hereditary polyposis syndrome, so they are often unsure of how to manage care appropriately. They want to know:

- When do I start testing? What kind?
- How do I manage this syndrome?
- When surgery is indicated, what kind?

To date there have been no published guidelines specifically for children at risk of or affected by familial adenomatous polyposis, Peutz-Jeghers syndrome or juvenile polyposis syndrome.

Thomas M. Attard, MD, pediatric gastroenterologist with the Polyposis Center at Children's Mercy Kansas City, has worked to change that. Dr. Attard is a member of ESPGHAN's Polyposis Working Group, an effort that brought together recognized experts in the subject from across the world and who worked diligently over six years to create clear recommendations to serve as practical guides for use with pediatric and adolescent patients. Formed when the European Society for Paediatric Gastroenterology, Hepatology and Nutrition (ESPGHAN) recognized the need for polyposis guidelines in 2012, the Polyposis Working Group reached final consensus in 2018. Guidelines were then presented, and position papers were published in the *Journal of Pediatric Gastroenterology and Nutrition* in March 2019.

The Polyposis Center at Children's Mercy is the only comprehensive, multidisciplinary team in the region with expertise in diagnosing and treating pediatric polyps and related intestinal cancer syndromes.

ACHIEVING CONSENSUS ON CARE

The new recommendations are based on committee members' review of existing literature and their expert opinions. They specifically address diagnosis, assessment, screening and treatment of all three hereditary polyposis syndromes in children and adolescents. With these new recommendations, gastroenterologists now have practical guidance for management of pediatric patients with these conditions.

The position papers support and educate providers to deliver informed, cautious care. Cancer risk is defined as clearly as can be, given the current evidence, and procedures and tests to prevent illness are tailored accordingly.

AN OVERVIEW OF THE NEW RECOMMENDATIONS

Focus on syndrome-specific targets:

- Familial adenomatous polyposis: Focus on the timing of surgery to remove the colon
- Peutz-Jeghers syndrome: Focus on avoiding polyps that cause intestinal obstruction
- Juvenile polyposis syndrome: Focus on the genetic subgroup because of differences in non-intestinal testing needs

When to perform genetic testing:

- Familial adenomatous polyposis: 12-14 years
- Peutz-Jeghers syndrome: 3 years
- Juvenile polyposis syndrome: 12-15 years

All genetic testing should be conducted in conjunction with genetic counseling.

AN OVERVIEW OF THE NEW RECOMMENDATIONS

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Who to test:

- Positive family history (autosomal dominant pattern of inheritance)
- Suspicious clinical findings:
 - Bilateral retinal abnormalities CHRPE (FAP)
 - Liver cancer, hepatoblastoma (FAP)*
 - Multiple benign desmoids (FAP)
 - Mucocutaneous freckling (PJS)
- Meet clinical criteria (endoscopic/extraintestinal findings)

*Although these are not officially part of the recommendations, they are in clinical practice today.

Diagnostic criteria:

For the first time, consensus exists among pediatric GI specialists about how to diagnose, as shown in the table below.

Polyposis Syndrome	Diagnostic Criteria
Juvenile Polyposis Syndrome	<ol style="list-style-type: none">1. Five or more JPS of the colon or rectum, OR2. JPS in other parts of the GI tract, OR3. Any number of JPS and a positive family history
Peutz-Jeghers Syndrome	<ol style="list-style-type: none">1. Two or more histologically confirmed PJS polyps, OR2. Any number of PJS polyps in an individual who also has characteristic mucocutaneous pigmentation, OR3. Any number of PJS polyps OR characteristic mucocutaneous pigmentation in an individual who has a family history of PJS in close relative(s)
Familial adenomatous polyposis	<ol style="list-style-type: none">1. Pathogenic APC mutation, OR2. Multiple colonic adenomas identified on endoscopy (at least 10-20 cumulative) with or without APC mutation, OR3. Child with a desmoid tumor with a known family history of FAP

Complete details on the clinical recommendations for all three hereditary polyposis syndromes are included in the ESPGHAN Polyposis Working Group position papers. Please visit transformpediatrics.childrensmercy.org/recommendations to learn more.

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LEARN MORE ABOUT THE POLYPOSIS CENTER AT CHILDREN'S MERCY.

Thomas M. Attard, MD
tmattard@cmh.edu • (816) 302-3028
transformpediatrics.childrensmercy.org



For consults, admissions or transport call: 1 (800) GO MERCY / 1 (800) 466-3729.

Winter 2020