Hereditary colon cancer syndrome marked by abnormally dense blood vessel growth in mouth

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A team led by Johns Hopkins researchers has found that a hereditary colon cancer syndrome, familial adenomatous polyposis (FAP), is associated with abnormally dense blood vessel growth in the skin lining the mouth.

The finding, reported in the June issue of *Familial Cancer*, could lead to a quick screening test for FAP, which is normally diagnosed with expensive DNA tests and colonoscopies, and sometimes goes unnoticed until cancer develops.

"This higher blood vessel density in the mouth may reflect an abnormal state of cells lining the digestive tract – including the oral cavity – that predisposes people to colorectal cancer and precancerous polyps," says Francis M. Giardiello, M.D., Johns G. Rangos Sr. Professor of Medicine at Johns Hopkins and director of Hopkins' Hereditary Colorectal Cancer Program.

People who have even one copy of the mutant gene that causes FAP develop hundreds of precancerous colorectal polyps, also known as adenomas, in their teens. Most have their colons removed after diagnosis to avoid what would otherwise be a near-100 percent risk of colon cancer by middle age.

In 2003, Italian researchers reported that a similar genetic condition,

hereditary non-polyposis colorectal cancer (HNPCC), was linked to a greater complexity of blood vessels in the oral mucosa – the skin that lines the mouth. Daniel L. Edelstein, a senior research program coordinator at Johns Hopkins University School of Medicine, says he read the Italian report and brought it to Giardiello's attention.

Edelstein also contacted Jessica C. Ramella-Roman, an expert on bio-optics systems at The Catholic University of America. "She developed a cameralike device that enabled a direct and relatively automated measurement of this vascular density in the lining of the mouth," he says.

Using Ramella-Roman's device and associated image-analysis software, the researchers scanned a two-centimeter-square patch of oral mucosa inside the lower lip of 33 patients with FAP. All 33 were enrolled in the Johns Hopkins Hereditary Colorectal Cancer Registry. The team also scanned a similar tissue sample of 50 control subjects who were matched for age and other variables but had no personal or family history of colorectal cancer or adenoma(s). Each subject was screened to determine the density of visible blood vessels in their lower lip – a measure they called the "oral mucosal vascular density (OMVD)."

"The OMVD measures were significantly higher in FAP patients than in healthy controls," says Giardiello. "About 90 percent of FAP patients in this sample had OMVD values above a certain threshold, and about 90 percent of controls were below that threshold, so in principle, we could use that threshold for screening purposes." Differences in the OMVD results were unrelated to age or gender, according to the researchers.

To further investigate the technique's screening potential, the researchers gave the OMVD test to five of Giardiello's patients who had multiple polyps but no detectable mutation for FAP or HNPCC on genetic tests. "They might have other, unknown gene mutations predisposing them to polyp formation, or they might have FAP or HNPCC mutations that

somehow weren't picked up in the tests," said Giardiello.

All five of these patients had OMVD scores above the high-risk threshold. "The results suggest that this high-OMVD condition may be an alternative marker for colon cancer risk, even when we can't find a gene mutation," Giardiello says.

Tumors typically promote the spread of new blood vessels in their vicinity to maintain their high growth rates. FAP mutations also boost the production of factors that increase new-vessel growth in the colon and other tissues. That could explain why people with FAP have higher vascular densities in their mouths, says Giardiello.

"While there seems to be a reason why FAP patients have this denser vessel growth, I don't yet have a plausible explanation for how HNPCC gene mutations could cause this overgrowth," says Giardiello. "It's something that we'd like to investigate further."

Provided by Johns Hopkins Medical Institutions

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