SY-18 Symposium
08:30—12:00 Hermès

Joint Session ESP & IARC
Classification of genetic tumour syndromes
*Chairs: Ian Cree, France*
*Fátima Carneiro, Portugal*

001 08:30—08:50
How the WHO Blue Books are handling hereditary cancer syndromes
*Ian Cree, France*

002 08:50—09:00
Overview of the digestive system syndromes
*Fátima Carneiro, Portugal*
Access to the following books:

5th edition
Digestive Tumours
Breast Tumours

4th edition
Skin Tumours
Eye Tumours
Endocrine Tumours
Head and Neck Tumours

WHO Classification of Tumours
ONLINE
Now available at: tumourclassification.iarc.who.int

Special launch rate of 100 Euros

More details from the IARC team – Booth A14, 2nd level, Agora 2 Hall
9am to 5.15pm, from Sunday 8 to Tuesday 10 September
### Genetic tumour syndromes of the digestive system

<table>
<thead>
<tr>
<th>Disease/phenotype</th>
<th>Syndrome MIM number</th>
<th>Inheritance</th>
<th>Locus</th>
<th>Gene(s)</th>
<th>Gene MIM number</th>
<th>Encoded protein(s)</th>
<th>Normal protein function</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>609310</td>
<td>AD</td>
<td>3p22.2</td>
<td>MLH1</td>
<td>120436</td>
<td>MLH1</td>
<td></td>
</tr>
<tr>
<td></td>
<td>120435</td>
<td>AD</td>
<td>2p21-</td>
<td>MSH2</td>
<td>609309</td>
<td>MSH2</td>
<td>DNA mismatch repair</td>
</tr>
<tr>
<td></td>
<td>614350</td>
<td>AD</td>
<td>2p16.3</td>
<td>MSH6</td>
<td>600678</td>
<td>MSH6</td>
<td></td>
</tr>
<tr>
<td>Lynch syndrome</td>
<td>614337</td>
<td>AD</td>
<td>7p22.1</td>
<td>PMS2</td>
<td>600259</td>
<td>PMS2</td>
<td>Calcium-independent cell–cell adhesion; mutation results in epigenetic silencing of MSH2</td>
</tr>
<tr>
<td></td>
<td>613244</td>
<td>AD</td>
<td>2p21</td>
<td>EPCAM</td>
<td>185535</td>
<td>EPCAM</td>
<td></td>
</tr>
</tbody>
</table>

- lists each of the syndromes discussed in this chapter
- summarizes key information about the disease/phenotype, pattern of inheritance, causative gene(s), and normal function of the encoded protein(s).

Some of the syndromes included in the table are not discussed in detail in this chapter because of space limitations.
### Genetic tumour syndromes of the digestive system

#### Common syndromes

<table>
<thead>
<tr>
<th>Disease/phenotype</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lynch syndrome</td>
<td>Common syndromes including Lynch syndrome and Familial adenomatous polyposis 1 (FAP) are covered in detail, as well as several other adenomatous polyposes defined since the last volume and the GAPPs (Gastric adenocarcinoma and proximal polyposis of the stomach) syndrome, now recognized as a FAP variant, with an unique phenotype.</td>
</tr>
<tr>
<td>Li–Fraumeni syndrome (LFS)</td>
<td>A number of other genetic tumour predisposition syndromes that confer a raised risk of various gastrointestinal tumours are also described, including Li–Fraumeni syndrome (LFS), hereditary haemorrhagic telangiectasia (HHT), syndromes associated with gastroenteropancreatic neuroendocrine tumours (NETs), and multilocus inherited neoplasia alleles syndrome (MINAS).</td>
</tr>
</tbody>
</table>

This should be helpful to many involved in the diagnosis of such syndromes, as well as those researching the mechanisms involved.
Diagnostic algorithm for patients with adenomatous polyposis in whom familial adenomatous polyposis (FAP) and Lynch syndrome have been excluded

- CMMRD, constitutional mismatch repair deficiency;
- MAP, MUTYH-associated polyposis
- NAP, NTHL1-associated polyposis
- PPAP, polymerase proofreading–associated polyposis
ENJOY THE CHAPTER ON GENETIC TUMOUR SYNDROMES OF THE DIGESTIVE SYSTEM

Thanks for your attention