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<td><strong>Author(s)</strong></td>
<td>Miao, X; GarciaBarceló, MM; So, MT; Leon, TYY; Lau, DKC; Liu, TT; Chan, EKW; Lan, LCL; Wong, KKY; Lui, VCH; Tam, PKH</td>
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Role of RET and PHOX2B gene polymorphisms in risk of Hirschsprung’s disease in Chinese population

Hirschsprung's disease (OMIM 142623) is a complex congenital disorder characterised by the absence of ganglion cells of the plexus myentericus and plexus submucosus in the distal part of the large bowel, resulting in failure to pass meconium, chronic severe constipation and colonic dilatation in the neonatal period. Aganglionosis is attributed to a defect of the enteric nervous system, in which ganglion cells are required for normal development of the enteric nervous system, is the major susceptibility gene for Hirschsprung’s disease. There is growing evidence indicating that functional single nucleotide polymorphisms (SNPs) of RET could act as low susceptibility factors for Hirschsprung’s disease. In addition, PHOX2B encodes a transcription factor that is involved in the development of the autonomic neural crest derivatives.

Interaction of the RET and PHOX2B genotypes combined with each other affects the risk of Hirschsprung’s disease. However, the essential mechanisms behind our finding need to be investigated.

Acknowledgements

We extend our gratitude to all subjects who participated in the study. This work was supported by the research grant HKU 7392/04M from the Hong Kong Research Grants Council to Maria-Merce Garcia-Barcelo.

Table 1 Risk of Hirschsprung’s disease associated with RET genotypes combined with PHOX2B genotypes

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Patients (n = 256)</th>
<th>Controls (n = 242)</th>
<th>OR* (95% CI)</th>
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</thead>
<tbody>
<tr>
<td>RET +/G/A</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>GG + GA</td>
<td>19 (7.4)</td>
<td>65 (26.9)</td>
<td>1.00</td>
</tr>
<tr>
<td>GG + AA</td>
<td>57 (22.2)</td>
<td>122 (50.4)</td>
<td>1.86 (1.20–2.75)</td>
</tr>
<tr>
<td>AA + GA</td>
<td>36 (14.1)</td>
<td>15 (6.2)</td>
<td>7.96 (1.64–17.60)</td>
</tr>
<tr>
<td>AA + AA</td>
<td>144 (56.3)</td>
<td>40 (16.5)</td>
<td>11.72 (6.35–21.55)</td>
</tr>
</tbody>
</table>

* ORs and 95% CIs were calculated by unconditional logistic regression adjusted for gender.

References


The incidence and risks of liver biopsy in non-cirrhotic patients: An evaluation of 3806 biopsies

Liver biopsy plays a crucial role in the diagnosis and management of liver diseases. For the past decade, this invasive procedure has become a safe one with the prevailing application of an ultrasound-guided method, the use of thinner gauge needles and improved operational techniques. Over the past years, the debatable issue of liver biopsy has mainly focused on the safety and suitability of a shorter observation time with respect to cost savings. Referring to this issue, Beddy et al even shortened their observation time to 1 hour and indicated that only one haemorrhagic complication occurred within one hour amongst 500 liver biopsy occasions. There were no recorded delayed complications or deaths at follow up.

Generally, we agree with the conclusion that outpatient liver biopsy is safe when done in a

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