

Genomic medicine: challenges and opportunities for physicians

H Burton, T Cole and AM Lucassen

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Errors were introduced to Table 1 in this article during typesetting. The ‘Genetically predisposed subgroup’ cell of the breast cancer row should read ‘About 5% of cancers will be largely due to inheritance of a mutation in *BRCA1* or *BRCA2*’, and the ‘Importance of molecular diagnosis in management’ cell of breast cancer should read ‘Further preventive management for breast cancer and/or ovarian cancer (eg mammography or MRI; prophylactic surgery of breasts and /or ovaries)’. The ‘Genetically predisposed subgroup’ cell of the bowel cancer row should read ‘About 5% of all cancers will be due, in large part, to inheritance of a mutated gene: eg familial polyposis, Lynch syndrome or *MutYH* polyposis’ and the ‘Importance of molecular diagnosis in management’ cell for bowel cancer should read ‘Consideration of other at-risk organs (eg uterus or ovaries in Lynch syndrome)’.

The corrected table is printed here.

Table 1. Common conditions and genetically predisposed subgroups.

Condition/disease/presentation to mainstream clinical specialist	Genetically predisposed subgroup	Underlying genetic variation	Importance of molecular diagnosis in management*
Arrhythmia/syncope/SCD	<ul style="list-style-type: none"> • Ion channelopathies (eg LQT syndrome) 	<ul style="list-style-type: none"> • Several different genes related to LQT syndrome that, if mutated, predispose to arrhythmias and SCD 	<ul style="list-style-type: none"> • Preventive treatments such as β blockers • Implantable defibrillator
Breast cancer	<ul style="list-style-type: none"> • About 5% of cancers will be largely due to inheritance of a mutation in <i>BRCA1</i> or <i>BRCA2</i> 	<ul style="list-style-type: none"> • <i>BRCA1</i> and <i>BRCA2</i> mutations 	<ul style="list-style-type: none"> • Further preventive management for breast cancer and/or ovarian cancer (eg mammography or MRI; prophylactic surgery of breasts and /or ovaries)
Bowel cancer	<ul style="list-style-type: none"> • About 5% of all cancers will be due, in large part, to inheritance of a mutated gene: eg familial polyposis, Lynch syndrome or <i>MutYH</i> polyposis 	<ul style="list-style-type: none"> • <i>APC</i> gene-related familial polyposis • Mismatch repair genes (eg <i>MSH2</i>, <i>Mlh1</i>) • Mutations in <i>MYH</i> (recessive inheritance) 	<ul style="list-style-type: none"> • Colonoscopic surveillance and removal of adenomas • Consideration of other at-risk organs (eg uterus or ovaries in Lynch syndrome) • Aspirin to reduce incidence of adenomas
Diabetes	<ul style="list-style-type: none"> • Maturity-onset diabetes in young people 	<ul style="list-style-type: none"> • Mutations in <i>MODY</i> gene 	<ul style="list-style-type: none"> • Respond to treatment with sulphonylurea, so insulin treatment may not be necessary
High cholesterol/premature CHD	<ul style="list-style-type: none"> • Familial hypercholesterolaemia • Young age of onset of high cholesterol 	<ul style="list-style-type: none"> • LDL receptor gene or apolipoprotein B gene 	<ul style="list-style-type: none"> • Treatment with statins, using high-intensity statins as necessary
Obesity	<ul style="list-style-type: none"> • Severe, early-onset obesity that may be associated with severe insulin resistance and other endocrinological disturbances 	<ul style="list-style-type: none"> • Genes involved in production of leptin, which is involved in regulation of appetite) and downstream pathways 	<ul style="list-style-type: none"> • Stringent restriction of access to food in patients with monogenic obesity • leptin treatment in some
Congenital deafness	<ul style="list-style-type: none"> • Usher’s syndrome 	<ul style="list-style-type: none"> • At least 11 different genetic loci; recessive condition, so both parents need to be carriers 	<ul style="list-style-type: none"> • Recognition of Usher’s syndrome is important, as the child will also lose vision and so should have early intervention to enhance communication (eg cochlear implantation)

CHD = coronary heart disease; LDL = low-density lipoprotein; LQT = long QT; MRI = magnetic resonance imaging; SCD = sudden cardiac death.

*Includes consideration and communication of risk to family members or future family members. Molecular diagnosis allows cascade testing to take place to identify affected individuals and can reassure those who have not inherited the familial predisposition that they do not need interventions.