

# Clinical Genetics Service

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Dear Sirs

**Response to document:**

**“A Healthier Future; A Twenty Year Vision for health and Wellbeing in Northern Ireland 2005 – 2025”**

I write to respond to your document “A Healthier Future; A Twenty Year Vision for Health and Wellbeing in Northern Ireland 2005 – 2025”. I was particularly concerned to read paragraph 2.12 where it mentioned that “some genetic tests are available now, principally for the purposes of pre-natal diagnosis and pregnancy advice and for informing various forms of treatment” and the subsequent paragraph 2.13 where it mentions “in the future genetic testing and screening may significantly shift to the point of intervention for many conditions where we currently can diagnose and treat only after the emergence of symptoms, in the future we will be able to intervene earlier and our interventions will increasingly be in the form of health promotion and protection activities”.

Paragraph 2.12 was possible in the early 1990’s as pre-natal diagnosis has been available since then. Over the last 5 years, around 50% of referrals to the Regional Genetic Service have been referrals to the Cancer Genetic Service within Regional Genetics. These allow us, particularly for the conditions of breast, bowel and ovarian cancer, to allow genetic testing to predict what a particular gene is in the family and as most of these genes are autosomal dominant 50% of people in families will be at risk of carrying a gene. This means that screening and prevention is already possible, e.g. if a lady aged 45 develops colon cancer and a gene is isolated and shows that she carries a mutation within the MSH2 gene, not only can we screen this lady for other related cancers including her risk of developing endometrial cancer of 40% and a risk of ovarian cancer of 10%, but we can also screen her children with 2-yearly colonoscopies from the ages of 25 right up to 60 and this will allow any polyps that develop in the bowel to be removed during the colonoscopy procedure and thus completely avoiding bowel cancer in that person without intervention of surgery or chemotherapy.


The risk for carrying such a gene and developing colon cancer is around 70%.

There are 2 limiting features in all of this. The first is a resource issue from the Regional Genetic Service. Currently an action plan is being formulated from the regional review of genetic services and more money will allow us to enhance this already cutting edge service. The second restriction is that screening for breast, colon and ovarian cancers that are hereditary require resources including mammography, colonoscopy and staff to screen ovaries. Even though we have regional guidelines that have been circulated and a revision has just been sent out to all GPs and relevant practitioners and this has immensely improved the number of patients coming through family history clinics in the cancer units led by colorectal, breast and ovarian surgeons, they are struggling to keep up with the recommended screening measures.

This means that the current managed clinical network is in place and works fairly well but needs more resources to ensure that the recommendations that are given following genetic testing in 10% of cancers can be implemented. If this worked fully then there would be large cost savings to the Health Service through prevention and avoidance of cancers in up to 10% of the population. This model can then be used for other polygenic disorders such as diabetes, heart disease and so on and these groups of patients and disorders are where you should be concentrating on with the 2025 vision.

I hope this is helpful.

Yours sincerely



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Consultant Clinical Geneticist**

Copy to Dr TCM Morris, Chairman, Radiotherapy & Oncology Sub-Committee,  
AMAC, Haematology Dept; Belfast City Hospital Trust

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