NEW ZEALAND MEDICAL ASSOCIATION

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Dear Martin

Development of a National Clinical Genetics Service

We are pleased to be given the opportunity to comment on this document, although are disappointed with the extremely short time frame provided.

The NZMA is New Zealand's largest medical organisation and has a pan professional membership. We have around 4000 members who come from all areas of medicine including medical students, resident medical officers, general practitioners, and other specialists.

The NZMA aims to provide leadership of the medical profession, and promote:

- professional unity and values;
- the health of New Zealanders.

The key roles of the NZMA are to:

- provide advocacy on behalf of doctors and their patients;
- provide support and services to members and their practices;
- publish and maintain the Code of Ethics for the profession; and
- publish the New Zealand Medical Journal.

In principal, the NZMA supports the proposed national co-ordination of genetic services and the need for appropriate funding. The critical issue is the provision of clinical genetic services into the future. This is another (sub) specialist area with an international shortage of appropriately trained practitioners.

However this draft document lacks focus and fails to differentiate important elements under the genetic term "Genetic Services".

There are two aspects to genetic services:-

1. Clinical genetic services, where genetic advice is provided to patients and families with rare genetic disorders by clinicians who have trained as clinical geneticists.

2. Laboratory genetic services.

Molecular testing is becoming a basic tool within laboratory medicine and genetic techniques underpin a number of laboratory tests. Molecular genetic testing is, however, primarily carried out within hospital reference laboratories.

Examples of tests performed by these reference laboratories include:-

- Molecular characterisation of acute leukaemias relevant to treatment and prognosis.
- Genetic testing for single gene disorders such as thalassaemia and haemophilia.
- Molecular markers for the familial predisposition to thrombotic diseases (thrombophilia testing).
- Somatic mutation analysis for familial cancer risk.
- Molecular diagnosis of myeloproliferative disorders.

Laboratory genetic services have largely been established by pathologists or clinician/pathologists (e.g. haematologists) in response to a clinical need within their own speciality or in response to an established need within another specialty (e.g. oncology). The expertise for interpreting and advising patients on the significance of these results largely resides within the specialty group that has spearheaded the development of the laboratory testing. Examples of this include haemophilia specialists providing patients under their care with genetic advice, or haematologists advising on the risks of a couple who carry the thalassaemia gene on the likelihood of having a child with a more severe genetic abnormality. Genetic counselling in this setting is most appropriately left within the expert group providing overall management of the disorder.

Under Section 8.3 (Page 49) *"Achieving Utilisation Management",* there is a suggestion that all funding will be taken from DHBs, which is currently used for genetic testing and giving it to a new genetic clinical service which will become the "gatekeeper" on the appropriateness of this testing. There is the potential that the testing may be refused. This suggests that clinical geneticists should inherit complete responsibility for the funding and scope of all molecular testing which is currently appropriately and easily accessed by a range of medical practitioners, including those in general practice (e.g. haemochromatosis testing; thrombophilia markers).

This would be inappropriate, as the expertise for performing this testing and interpreting the results resides within the subspecialty groups who have been involved in establishing the testing in the first instance. In this case, the molecular

test results are just another part of the clinical and routine laboratory jigsaw. Clinical geneticists cannot be assumed to have special expertise in this area.

Further, we believe that there is little to be gained by arbitrarily changing funding streams and allocating this responsibility to a “National Genetics Service”. Rather than looking to develop a central genetic provider, given that many specialists have considerable knowledge regarding the management of genetic disorders of patients within their discipline, consideration needs to be given to acknowledging the skills in these disciplines and looking to coordinate and acknowledge their role within the provision of an overall genetic services plan.

Yours faithfully

Dr Peter Foley
Chair