Wellington Regional Genetics Laboratory

March 2016

# Duchenne Muscular Dystrophy (DMD) and Becker Muscular Dystrophy (BMD) Disease Testing

Wellington Regional Genetics Laboratory (WRGL) is pleased to now offer DMD/BMD disease testing. Referrals for diagnostic queries can be accepted from neurologists, paediatricians and clinical geneticists.

## Background

Deletions and duplications of the *DMD* gene are frequent causes of DMD/BMD. Deletions account for 60-70% and duplications about for 5-10% of cases. Detection of these imbalances are routinely used in diagnostic testing of individuals symptomatic or those that are at risk of being carriers. DMD has a prevalence of 2.9 per 10,000 male births and BMD has a prevalence of 0.5 per 10,000 male births.

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## Testing at WRGL

Multiplex ligation-dependent probe amplification (MLPA) using the two commercially available probe sets (P034-B1 and P035-B2) is used to determine if there is a deletion or duplication present. The MLPA product can be visualised using fragment analysis on the 3130 Genetic Analyser. Results are compared with appropriate controls. A small proportion of cases will require further testing which is not offered at WRGL and samples will be forwarded to another laboratory if additional testing is required/necessary.

## Pricing, Sample Requirments and Turnaround Times

| Test | Cost NZD | Sample requirements | Turnaround time |
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| DMD/BMD | $284.20 | 2 x 3 ml of whole blood in EDTA (purple top) | 2 weeks<br>For urgent cases please contact the laboratory |

*****No direct charge for the central region DHBs covered by the Crown Funding Agreement

If you require any additional information about this test or any other aspect of our service, please do not hesitate to contact us.

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