MOTOR NEURONE DISEASE
Produced by the Centre for Genetics Education. Internet: http://www.genetics.edu.au

MND.

Includes: amyotrophic lateral sclerosis (ALS; Lou Gehrig’s disease), bulbar palsy, progressive muscular atrophy, Werdnig-Hoffman disease (infantile spinal muscular atrophy), Kugelberg-Welander syndrome (juvenile spinal muscular atrophy), and benign focal amyotrophy

FACTS

Motor neurone disease or ALS is characterised by a progressive muscle weakness and wasting, which affects movement of the limbs, speech, swallowing and respiration. In the majority of cases, this is not associated with intellectual impairment. Approximately 10-15 % of cases are thought to be inherited. The remainder are sporadic (affecting only one member of the family) and the cause is unknown.

A number of forms of inherited or familial motor neurone disease have been identified. There may be incomplete penetrance, where not all individuals carrying a faulty gene causing motor neurone disease, will necessarily develop the condition. In some families where the condition is inherited, mutations have been identified in a gene called the SOD1, which is located on chromosome 21. In these cases, the inheritance of the faulty gene follows a pattern of inheritance called autosomal dominant inheritance. Other causative genes include VAP8 and ALS2.

Progressive bulbar palsy has an autosomal recessive pattern of inheritance. Werdnig-Hoffmann disease (spinal muscular atrophy type I) is transmitted as an autosomal recessive condition and the SMN1 and SMN2 genes causing this condition are located on chromosome 5. Kugelberg-Welander syndrome is an autosomal dominant condition. Benign focal amyotrophy and infantile spinal muscular atrophy are inherited as autosomal dominant conditions.

Support Groups can provide individuals and families with information about genetic conditions, community resources and an understanding and empathic ear.

If you are not able to contact the services listed below, please call the Association of Genetic Support of Australasia (AGSA) on (02) 9211 1462 or visit www.agsa-geneticsupport.org.au for support and assistance.

Further information and details of your local genetic counselling service can be found at www.genetics.edu.au, or please call The Centre for Genetics Education on (02) 9462 9599.