Australian Genomics

Differential diagnoses
(please give multiple diagnoses, multiple diagnoses help the laboratory identify the disease gene)
Symptoms/ findings for referral
(please give main points here and/or attach a clinic letter)
Age of onset
☐ Congenital ☐ Juvenile (5-15 years) ☐ Infantile (<1 year) ☐ Early adult (16-40 years) ☐ Childhood (1-5 years) ☐ Late adult (>40 years)
Evolution of symptoms
☐ Rapidly progressive ☐ Slowly progressive ☐ Non-progressive
Previous genotype data
Previous single gene test/s
e.g., *SMN1*, *DMD*
Concluded clinical phenotype

Phenotype			
a. Muscle weakness	☐ Yes	☐ No	☐ Not assessed
i. Facial	☐ Yes	☐ No	
ii. Ptosis	☐ Yes	☐ No	
iii. Ophthalmoplegia	☐ Yes	☐ No	
iv. Axial	☐ Yes	☐ No	
v. Prox. UL	☐ Yes	☐ No	
vi. Prox. LL	☐ Yes	☐ No	
vii. Distal UL	☐ Yes	☐ No	
viii. Distal LL	☐ Yes	☐ No	
i. Facial	☐ Mild	☐ Moderate	☐ Severe
ii. Ptosis	☐ Mild	☐ Moderate	☐ Severe
iii. Ophthalmoplegia	☐ Mild	☐ Moderate	☐ Severe
iv. Axial	☐ Mild	☐ Moderate	☐ Severe
v. Prox. UL	☐ Mild	☐ Moderate	☐ Severe
vi. Prox. LL	☐ Mild	☐ Moderate	☐ Severe
vii. Distal UL	☐ Mild	☐ Moderate	☐ Severe
viii. Distal LL	☐ Mild	☐ Moderate	☐ Severe
ix. Short comment:			
b. Muscle atrophy/hypotrophy	☐ Yes	☐ No	☐ Not assessed
i. Facial	☐ Yes	☐ No	iv. Prox. LL ☐ Yes ☐ No
ii. Axial	☐ Yes	☐ No	v. Distal UL ☐ Yes ☐ No
iii. Prox. UL	☐ Yes	☐ No	vi. Distal LL ☐ Yes ☐ No
vii. Short comment:			
c. Muscle hypertrophy	☐ Yes	☐ No	☐ Not assessed

i. Calf ☐ Yes ☐ No
ii. Tongue ☐ Yes ☐ No
iii. Brachioradialis ☐ Yes ☐ No
iv. Other:

d. Myotonia ☐ Yes ☐ No ☐ Not Assessed
i. Chronic ☐ Yes ☐ No
ii. Provoked ☐ Yes ☐ No
Affected muscles:

e. Muscle stiffness ☐ Yes ☐ No ☐ Not assessed
f. Muscle pain ☐ Yes ☐ No ☐ Not assessed
g. Muscle cramps ☐ Yes ☐ No ☐ Not assessed
h. Muscle fatigue ☐ Yes ☐ No ☐ Not assessed
i. Exercise intolerance ☐ Yes ☐ No ☐ Not assessed
j. Fasciculations ☐ Yes ☐ No ☐ Not assessed
k. Rhabdomyolysis ☐ Yes ☐ No ☐ Not assessed
l. Malignant hyperthermia ☐ Yes ☐ No ☐ Not assessed
Comments (f-l):

m. Respiratory distress ☐ Yes ☐ No ☐ Not assessed
i. Assisted ventilation ☐ Continuous ☐ Intermittent ☐ Invasive ☐ Non-invasive

n. Joint hyperlaxity/hypermobility ☐ Yes ☐ No ☐ Not assessed
i. Neck ☐ Yes ☐ No
ii. Elbows ☐ Yes ☐ No
iii. Fingers ☐ Yes ☐ No
iv. Hip ☐ Yes ☐ No
v. Knees ☐ Yes ☐ No
vi. Ankles ☐ Yes ☐ No
vii. Toes ☐ Yes ☐ No
vii. Beighton score: _____/9

o. Joint contractures ☐ Yes ☐ No ☐ Not assessed
i. Arthrogryposis multiplex congenita ☐ Yes ☐ No
ii. Neck ☐ Yes ☐ No
iii. Elbows ☐ Yes ☐ No
iv. Fingers ☐ Yes ☐ No
v. Hip ☐ Yes ☐ No
vi. Knees ☐ Yes ☐ No
vii. Ankles ☐ Yes ☐ No

p. Bulbar dysfunction ☐ Yes ☐ No ☐ Not assessed
i. Dysarthria ☐ Yes ☐ No
ii. Dysphagia ☐ Yes ☐ No
iii. Dysarthria + dysphagia ☐ Yes ☐ No
iv. Feeding: ☐ NGT ☐ Gastrostomy

q. Deep tendon reflexes ☐ Yes ☐ No ☐ Not assessed

	LEFT			RIGHT		
i. Biceps	☐ Hypo	☐ Normal	☐ Hyper	Hyper ☐	Hypo ☐	Normal ☐
ii. Triceps	☐ Hypo	☐ Normal	☐ Hyper	Hyper ☐	Hypo ☐	Normal ☐
iii. Patellar	☐ Hypo	☐ Normal	☐ Hyper	Hyper ☐	Hypo ☐	Normal ☐
iv. Ankle	☐ Hypo	☐ Normal	☐ Hyper	Hyper ☐	Hypo ☐	Normal ☐

r. Skeletal deformities ☐ Yes ☐ No ☐ Not assessed

i. Scoliosis ☐ Yes ☐ No
ii. Lordosis ☐ Yes ☐ No
iii. Kyphosis ☐ Yes ☐ No
iv. Pectus excavatum ☐ Yes ☐ No
v. Pectus carinatum ☐ Yes ☐ No

s. Cardiac abnormalities ☐ Yes ☐ No ☐ Not Assessed
i. Cardiomyopathy ☐ Yes ☐ No ☐ Not Assessed
ii. Conduction defects ☐ Yes ☐ No ☐ Not Assessed

t. Walking ☐ Not Assessed

☐ Normal
☐ Can't jump
☐ Can't run
☐ Can't hop
☐ Can't manage stairs
☐ Gowers manoeuvre
☐ Walks with assistance (mobility aid walker)
☐ Fatigability
☐ Non-ambulant: intermittent
☐ Non-ambulant: permanent
i. Age of loss of ambulation: _____ yrs

Prenatal/Neonatal

a. Prenatal

i. Pregnancy ☐ Normal ☐ Abnormal ☐ Unknown
a) Foetal movements ☐ Normal ☐ Abnormal ☐ Unknown
b) Foetal dysmorphology ☐ Yes ☐ No ☐ Unknown
c) Polyhydramnios ☐ Yes ☐ No ☐ Unknown
ii. Labour ☐ NVD ☐ Abnormal ☐ Unknown
☐ Vacuum
☐ Forceps
☐ Caesarean
☐ Other

a) Preterm (<36 weeks) ☐ Yes ☐ No ☐ Unknown
If Yes, weeks gestation:_____

Specify:

b. Neonatal

i. Ventilatory support ☐ Yes ☐ No
a) If "Yes", has it improved over time? ☐ Yes ☐ No
ii. Supported feeding, e.g., NGT ☐ Yes ☐ No
b) If "Yes", has it improved over time? ☐ Yes ☐ No
iii. Delayed head/neck control ☐ Yes ☐ No
iv. Delayed early motor milestones ☐ Yes ☐ No

Comment: CNS involvement (e.g., learning difficulties, MRI brain), dysmorphic facial features, etc.

Findings on laboratory examinations

(describe significant findings)

a. Serum CK values: _____ U/L Age when CK tested:
Normal range for laboratory used: _____ U/L Date of CK test:

b. Electromyography (EMG) ☐ Assessed ☐ Not Assessed

- i. Myopathic ☐ Yes ☐ No
- ii. Mixed (myo/neuro) ☐ Yes ☐ No
- iii. Spontaneous activity ☐ Yes ☐ No
- v. Neurogenic ☐ Yes ☐ No
- vi. Myotonic discharges ☐ Yes ☐ No
- vii. EMG comment:

c. Nerve conduction studies ☐ Assessed ☐ Not Assessed

- ☐ Demyelinating
- ☐ Axonal
- ☐ Mixed
- ☐ Normal

UL motor conduction velocity: _____ m/s
ii. Short comment:

d. Nerve biopsy

- i. ☐ Normal ☐ Abnormal ☐ Not Assessed
- ii. Short comment:

e. MRI results

- i. Brain ☐ Normal ☐ Abnormal ☐ Not Assessed
- ii. Muscle ☐ Normal ☐ Abnormal ☐ Not Assessed
- iii. Other MRI:
- iv. Short comment:

f. Muscle biopsy ☐ Not Assessed

- i. Dystrophic ☐ Yes ☐ No
- ii. Myopathic ☐ Yes ☐ No
- iii. Neurogenic ☐ Yes ☐ No
- iv. Minor pathology ☐ Yes ☐ No
- v. Rimmed vacuoles ☐ Yes ☐ No
- vi. Myofibrillar pathology ☐ Yes ☐ No
- vii. Cores-multiminicores ☐ Yes ☐ No
- viii. Nemaline bodies ☐ Yes ☐ No
- ix. Central bodies ☐ Yes ☐ No
- x. >30% fibres with internal nuclei ☐ Yes ☐ No
- xi. Lymphocyte infiltrates ☐ Yes ☐ No
- xii. Secondary Calpain3 defect ☐ Yes ☐ No
- xiii. Normal ☐ Yes ☐ No
- xiv. Endstage ☐ Yes ☐ No
- xv. Short comment:

g. Fibro-adipose replacement of muscle ☐ No ☐ MRI ☐ CT ☐ Not Assessed

- i. Gluteal ☐ Yes ☐ No
- ii. Obturator ☐ Yes ☐ No
- vi. Calf ☐ Yes ☐ No
- vii. Anterior lower leg ☐ Yes ☐ No

iii. Quads	☐ Yes	☐ No	viii. Peroneal	☐ Yes	☐ No
iv. Thigh adductors	☐ Yes	☐ No	ix. Paravertebral	☐ Yes	☐ No
v. Hamstrings	☐ Yes	☐ No	x. Shoulders	☐ Yes	☐ No

xi. Specify as needed:

xii. Short comment:

h. Spirometry ☐ Yes ☐ Not assessed

i. FEV_1 values:_____
ii. FVC values:_____
iii. FEV_1/FVC ratio:_____

i. Echocardiography

i. ☐ Yes- normal ECG ☐ Yes- abnormal ECG ☐ Not assessed
ii. Abnormality:

General comments