

Mitochondrial Disorders Clinical Impact of Result

Patient: [Patient name prefilled]

Treating clinician: [Clinician name prefilled]

Genomic testing result:

Result: [Result prefilled: positive, negative or VOUS]

Pathogenic/likely pathogenic genes: [Gene(s) prefilled]

SECTION I Changes in patient management arising from test result

Please tick all relevant changes in patient management arising from the test result (whether positive, negative or VOUS).

1. Change in diagnostic investigations

- additional investigation initiated
- unnecessary investigation cancelled or prevented

2. Change in pharmaceutical and other treatments

Change in specific treatment for underlying cause (whether treatment started or unnecessary treatment stopped)

- specific treatment started
- unnecessary specific treatment stopped or not initiated

Change in nonspecific treatment for underlying cause

- nonspecific treatment started
- unnecessary nonspecific treatment stopped or not initiated

Change in treatment for other aspects of the condition (e.g. movement disorder, gastrointestinal symptoms, other neurological symptoms, other organ problems)

- treatment for other aspects of the condition started
- unnecessary treatment for other aspects of the condition stopped or not initiated

3. Change in other clinical management

- Change in screening for comorbidities
 - screening for comorbidities initiated
 - screening for comorbidities cancelled
- Change in referral to subspecialist
 - subspecialist referral initiated
 - subspecialist referral no longer required
- Change in overall direction of care
 - patient has been redirected towards palliation as a result of testing
 - previous decision to palliation has been reversed

SECTION II Overall impact of test result

How do you rate the clinical utility of genomic testing, in terms of changing process of management, for this patient?

- Not useful
- Neutral
- Useful
- Very useful

How do you rate the clinical utility of genomic testing, in terms of improving health outcomes, for this patient?

- Not useful
- Neutral
- Useful
- Very useful

Please specify the details on change in patient management arising from the test result that you selected in the previous section [Note: Only the pre-selected aspects of change will pop out; Clinicians will have chance to re-select or de-select the relevant fields]

SECTION I Changes in patient management arising from test result (either positive, negative or VOUS)

1. How did test result change diagnostic investigations for aetiology?

Additional investigation(s) ordered to clarify the diagnosis

Please specify the additional investigation(s) ordered

<input type="checkbox"/> Specific biochemistry: Name of the test(s) _____ [+] add new field if required
<input type="checkbox"/> MRI/MRS
<input type="checkbox"/> Biopsies/histology: <input type="checkbox"/> Nerve biopsy <input type="checkbox"/> Muscle biopsy <input type="checkbox"/> Liver biopsy <input type="checkbox"/> Others _____
<input type="checkbox"/> MRC enzymology
<input type="checkbox"/> Molecular genetic testing Name of the test(s) _____ [+] add new field if required

Unnecessary investigations cancelled or prevented

Please specify the investigations cancelled or prevented

<input type="checkbox"/> Specific biochemistry: Name of the test(s) _____ [+] add new field if required
<input type="checkbox"/> MRI/MRS
<input type="checkbox"/> Biopsies/histology: <input type="checkbox"/> Nerve biopsy <input type="checkbox"/> Muscle biopsy <input type="checkbox"/> Liver biopsy <input type="checkbox"/> Others _____
<input type="checkbox"/> MRC enzymology
<input type="checkbox"/> Molecular genetic testing Name of the test(s) _____ [+] add new field if required

2. How did test result change pharmaceutical and other treatments?

2.1 Allow specific treatment for underlying cause

Specific treatment started as a result of testing

Please specify the treatment(s) initiated

Name of treatment: _____ [+] add new field if required

Unnecessary specific treatment stopped or not initiated as a result of the testing

Please specify the treatment(s) stopped

Name of treatment: _____ [+] add new field if required

2.2 Allow nonspecific treatment for underlying cause

Nonspecific treatment started as a result of testing

Please specify the treatment(s) initiated

Name of treatment: _____ [+] add new field if required

Unnecessary nonspecific treatment stopped or not initiated as a result of testing

Please specify the treatment(s) stopped

Name of treatment: _____ [+] add new field if required

2.3 Change in treatment for other aspects of the condition (eg. movement disorder, gastrointestinal symptoms, other neurological symptoms, other organ problems)

Treatment for other aspects of the condition started

Aspect of condition 1 _____

Please specify the treatment(s) initiated

Name of treatment: _____ [+] add new field if required

[+] Add additional aspect of condition

Treatment for other aspects of the condition stopped

Aspect of condition 1 _____

Please specify the treatment(s) stopped

Name of treatment: _____ [+] add new field if required

[+] Add additional aspect of condition

3. How did test result change other clinical management?

3.1 Change in screening for comorbidities

Screening for comorbidities has been initiated

Comorbidity 1 _____

Please specify the screening initiated

Screening test: Frequency: Duration (number of years):
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[+] Add additional comorbidity

Screening for comorbidities has been cancelled

Comorbidity 1 _____

Please specify the screening cancelled

Screening test:
Frequency:
Duration (number of years):

[+] Add additional comorbidity

3.2 Change in referral to subspecialist

Subspecialist referral has been initiated

Please specify the subspecialty referred to and the frequency of visits

Subspecialty: _____
Number of visit(s) planned:
<input type="checkbox"/> One appointment
<input type="checkbox"/> Ongoing involvement of subspecialty planned
If ongoing visits planned, please specify:
Frequency of visits per year: _____
Planned duration of surveillance/treatment (number of years): _____

[+] add additional subspecialty if required

Prior subspecialist service is no longer required?

Please specify the subspecialty and the previously planned frequency of visits

Subspecialty: _____
Number of visit(s) planned:
<input type="checkbox"/> One appointment
<input type="checkbox"/> Ongoing involvement of subspecialty planned
If ongoing visits planned, please specify:
Frequency of visits per year: _____
Planned duration of surveillance/treatment (number of years): _____

[+] add additional subspecialty if required

3.3 Change in the overall direction of care

Patient has been redirected towards palliation as a result of testing

Comments _____

Previous decision to palliation has been reversed

Comments _____

SECTION II Overall impact of test result

[Note: Clinicians will have chance to review their answers and make changes if necessary.]

How do you rate the clinical utility of genomic testing, in terms of changing process of management, for this patient?

Not useful

Neutral

Useful

Very useful

How do you rate the clinical utility of genomic testing, in terms of improving health outcomes, for this patient?

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Neutral

Useful

Very useful