

## UKGTN Testing Criteria

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| <b>Test name:</b><br>GATA2 Deficiency  |  |
|  |  |
| <b>Approved name and symbol of disorder/condition(s):</b><br>Immunodeficiency 21; IMD21<br>Lymphedema, Primary, with Myelodysplasia<br>Leukemia, Acute Myeloid; AML<br>Myelodysplastic Syndrome; MDS | <b>OMIM number(s):</b><br>614172<br>614038<br>601626<br>614286 |
| <b>Approved name and symbol of gene(s):</b><br>GATA binding protein 2; GATA2   | <b>OMIM number(s):</b><br>137295                               |

|                          |                       |
|--------------------------|-----------------------|
| <b>Patient name:</b>     | <b>Date of birth:</b> |
| <b>Patient postcode:</b> | <b>NHS number:</b>    |
| <b>Name of referrer:</b> |                       |
| <b>Title/Position:</b>   | <b>Lab ID:</b>        |

| <b>Referrals will only be accepted from one of the following:</b> |                                    |
|---|------------------------------------|
| <b>Referrer</b>   | <b>Tick if this refers to you.</b> |
| Consultant Haematologist  |                                    |
| Consultant Clinical Geneticist                                    |                                    |
| Consultant Oncologist   |                                    |

| <b>Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:</b>   |  |
|---|--|
| <b>Criteria</b>   | <b>Tick if this patient meets criteria</b> |
| Recurrent, unexplained severe or prolonged mycobacterial, HPV, EBV or fungal infections or myelodysplastic syndrome <b>AND</b> at least one of the following: |  |
| Lymphocyte subset analysis showing reduced numbers of NK cells ± B cells  |  |
| Primary lymphedema  |  |
| Sensorineural deafness  |  |
| Monocytopenia   |  |
| Evidence of familial MDS/AML  |  |
| <b>OR</b> At risk family members where familial mutation is known.  |  |

### Additional Information:

If the sample does not fulfil the clinical criteria or you are not one of the specified types of referrer and you still feel that testing should be performed please contact the laboratory to discuss testing of the sample.