# ‘In the Clinic’ (Tier 1) template document

## **Presentation: A child with an intellectual disability**

**For some children presenting with an intellectual disability with developmental delay, there will be a genetic cause.**

**Title and summary:** For a presentation/testing-stage article, the title should describe the clinical presentation. (For a results-stage article, the title should describe the results, eg ‘Patient with colorectal cancer (germline DPYD variant)’.)   
The one-sentence summary below the title should highlight key point(s) of the article.

(Note the National Genomic Test Directory is organised mainly by condition, but we believe it is more helpful for this resource to be accessed by clinical presentation.)

The title is followed by a **one sentence strapline** to give context/summarise key points

**Example clinical scenario**

**Example clinical scenario:** This should describe a patient scenario that a clinician might encounter with this presentation. Keep it brief: 4-5 lines or 80 word maximum.

A family attend clinic concerned because their six-year-old son’s development is delayed: he sat at one year, was walking at 2.5 years and, at the age of six, has some single words but is not talking in sentences. He has some dysmorphic features and was diagnosed with an atrial septal defect following the detection of a heart murmur at the newborn check.

**When to consider genetic testing**

**When to consider genetic testing:** This should help to guide the clinician as to when genetic testing is appropriate for the presentation. The [test directory eligibility criteria document](https://www.england.nhs.uk/publication/national-genomic-test-directories/) has a list of testing criteria. If available, these criteria should be used here.

* Moderate to profound intellectual disability
* An intellectual disability (of any severity) associated with:
  + behavioural problems, including autistic spectrum disorder;
  + other medical problems, such as seizures, congenital heart disease;
  + abnormal growth patterns (growth retardation, overgrowth, asymmetric growth);
  + microcephaly or macrocephaly;
  + dysmorphic facial features; and/or
  + a family history of learning disability (particularly if X-linked pattern) or of multiple miscarriages

**What do you need to do?**

**What do you need to do:** This section is not about the diagnostic process or general management, but what you need to do in terms of ordering genomic testing.

* Consult the [test directory eligibility criteria](https://www.england.nhs.uk/publication/national-genomic-test-directories/) to ensure your patient is eligible for testing and to access a spreadsheet of all available tests.

Always include the first bullet point with links to test directory eligibility criteria and spreadsheet.

* Decide which of the panels best suits the needs of your patient/family. For developmental disorders, there are a number of available panels including:
  + R27: if you have already done array CGH and Fragile X testing and would like to investigate single gene causes of a child’s developmental delay/intellectual disability.
  + R29: if no genetic testing has yet been undertaken in a child with developmental delay/intellectual disability. This panel includes microarray, fragile X testing and sequencing.
  + R377: if only a microarray is required.
  + R47: if you think your patient might have a diagnosis of Angelman syndrome.
  + R48: if you think your patient might have a diagnosis of Prader Willi syndrome.
  + R53: if you think your patient might have a diagnosis of Fragile X syndrome.

**Conditions highlighted in green:** Make a note if any of the conditions you list are not currently included in the Knowledge Hub and need developing. Of note, the GEP’s [genetic condition factsheets](https://www.genomicseducation.hee.nhs.uk/doc-type/genetic-conditions/) can be adapted for this purpose (where available).

* A record of discussion (RoD) form is required. If you have not completed a RoD form before or do not have access to one, please find information here.
* Depending on the details you provide and the panel chosen, a range of genomic investigation techniques will be applied to your patient’s and, if appropriate, their family’s DNA. These include (but are not restricted to):
  + Whole genome sequencing
  + Whole exome sequencing
  + Gene panel
  + Single gene testing
  + Methylation studies
  + Southern blotting
  + Common aneuploidy testing
  + Microarray
  + MLPA

**Technologies highlighted in green:** Please prune as necessary. Each of these will link to a Tier 2 Technologies document, which will be available to link to as necessary.

* For DNA-based tests (all the above listed tests), an EDTA sample is required. For many of the tests (particularly whole genome and exome sequencing), parental samples are also needed/helpful. Please refer to your local GLH for details of test request forms and where to send samples.

**Resources for clinicians:**

**Resources for clinicians:** Link to any resources you think would be helpful (such as review papers, NICE guidelines, criteria for diagnosis, management guidelines and so on). Also there will be some printable Patient Communication Aids that might be relevant, such as for explaining AD/AR/XL inheritance. These are listed on the spreadsheet. Please also include the NGTD links as standard.

* [National Genomic Test Directory](https://www.england.nhs.uk/publication/national-genomic-test-directories/) and eligibility criteria

**Resources for patients:**

**Resources for patients:** Link to any recommended patient information. leaflets/support groups etc.