

Freedom of Information Act 2000 - Request Reference Fol/21/073
Downs Syndrome – Screening/Diagnostic Results

Request details

I wish to make a Freedom of Information request to ascertain the **language/terminology** used when **screening/diagnostic results** for **Down Syndrome** are given to **expectant parents**.

When results can be provided verbally and/or written, please provide content of each:

- Those given in a written format eg. letter/email
- Those given verbally - kindly provide any guidelines, templates or scripts used or referenced.

Please also indicate if you do not report such results directly, for example if another provider is responsible.

If different hospitals/maternity units within your board use different correspondence, please send examples from each unit, identifying the name of each hospital/maternity unit.

Please send an example of all the following that are in use;

- a) Copies of written materials/information provided to expectant women appertaining to antenatal screening test for Down Syndrome including information about both the tests and Down Syndrome.
- b) Templates reporting low chance combined or quadruple screen results
- c) Templates reporting high chance combined or quadruple screen results
- d) Templates reporting low chance NIPT results
- e) Templates reporting high chance NIPT results
- f) Templates reporting 'no result' NIPT results
- g) Templates reporting negative QF-PCR/FISH CVS results
- h) Templates reporting positive QF-PCR/FISH CVS results
- i) Templates reporting negative full karyotype CVS results
- j) Templates reporting positive full karyotype CVS results
- k) Templates reporting negative amniocentesis result
- l) Templates reporting positive amniocentesis result

Response details

Please find attached templates.

Antenatal Screening and available written information

It is important to highlight that when women are counselled they are offered screening tests not specifically for Down syndrome on its own, there are two tests that are offered. Within Wales, women if they choose to have the combined test cannot opt for one condition only the test will be for the three conditions.

Women are either provided with the Antenatal Screening Wales screening pack, or community midwives will offer to post the pack out especially in view of the current pandemic or the community midwife will signpost women to the following

<https://phw.nhs.wales/services-and-teams/screening/antenatal-screening-wales/antenatal-screening-tests/antenatal-screening-test-leaflet/>.

The information is also signposted on the CAV website that women can also access.

<https://cavuhb.nhs.wales/our-services/maternity-services/pregnancy/antenatal-resources/>.

The Combined screening test offers screening for Down Syndrome, Edwards syndrome and Patau's syndrome, if the woman consents to the screening it is for the three chromosomal conditions, women are unable to choose for example just screening of one of the conditions. The combined test is offered until 14+1 week's gestation or if the Crown rump length (CRL) is below 84mm.

The second test that would be offered would be the Quad test available from 15-18 weeks gestation, which again is a screening test with a slightly lower accuracy than the combined test and this test is only for Down syndrome screening.

Training updates for staff

- Midwives have access to the CAVUHB guideline on antenatal screening as well as being able to access ESR training on the three chromosomal conditions.
- Midwives are aware that screening can be emotive for many individuals and it is important to know that they are screening tests and not diagnostic but also there are pathways to follow dependent upon if the woman has a higher or lower chance result.
- Terminology such as "I am sorry to tell you" is not used by staff if there is a high chance result. The result is provided clearly and options discussed for further testing or no further testing. Staff are aware that a decision that may be right for one woman, is not necessarily right for another. Identifying pathways and Antenatal Screening Wales literature is also provided or signposted on the ASW website.

Template for reporting a low chance Combined or Quad test result:

The template letter is attached that is then sent out to the woman who receives a low chance result following Quad screening. The Woman's report is then married up with the letter and posted to the patient.

Template for reporting high chance Combined or Quad test result:

Women who have a higher chance result are telephoned by either the Screening Coordinator or midwife in Day assessment Unit who is confident and competent at giving results and discussing pathways that are available for the woman. Women are initially contacted and results are discussed, re-iterating that the result is based upon a screening test.

Options are provided for

- No further Screening
- Non- invasive prenatal testing (NIPT)
- Invasive/Diagnostic Testing

The CAVUHB pathway is followed and staff are aware that offering appropriate counselling and support and ensuring the woman has time to decide what is the best option for her and understanding that she can discontinue the screening pathway at any time. The woman will also be offered an appointment to attend and the consultation will go through the options available, what the options entail and provide written information. This is a booklet written by Antenatal Screening Wales that is provided to the woman; Information for women offered further tests for suspected chromosomal conditions.

<https://phw.nhs.wales/services-and-teams/screening/antenatal-screening-wales/what-do-results-mean/sickle-cell-and-thalassaemia/womens-chromosomal-conditions-booklet/>

Midwives will also sign post to Antenatal Results and Choices (ARC) <https://www.arc-uk.org/>

Low Chance NIPT

The Screening Coordinator or one of the midwives from Day assessment unit will contact the woman and inform of the result being low chance. This would be low chance for Down syndrome, Edwards's syndrome and also Patau's syndrome. The advice would be that the baby does not appear to be affected by any of the three chromosomal conditions but to advise that this was also not a diagnostic test. The woman would then be advised to continue with her planned antenatal care.

High Chance NIPT

The Screening coordinator or one of the experienced midwives in antenatal clinic will ring the woman with the result and advise of the higher chance result. The accuracy of the test is discussed and the Midwife will be sensitive and answer any questions that the woman may have. The woman will be offered referral to Fetal Medicine for Diagnostic / Invasive testing and there will be a discussion of the procedure and associate miscarriage risk. The woman is

offered time to consider her options. If in agreement a referral is made to the Fetal medicine department.

NIPT failed to report a result

This does happen occasionally and the woman is informed that there is not a high or low chance result. Therefore we are able to offer referral to Fetal Medicine Department for diagnostic/ Invasive testing in view that there is no NIPT result. The woman is advised that when this happens they do not repeat the NIPT test.

Results following Invasive/ Diagnostic Testing in Fetal Medicine Unit

Results within the department are accessed on viewpoint, a system that all fetal medicine departments use. Regardless of whether the woman has a confirmatory result for either T21, T18 or T13 or a normal result the results are telephoned through by the specialist midwives with in the FMU department.

Prior to the invasive testing, the woman is counselled by the specialist midwife as well as the Consultant to discuss the procedure and also the timing of results and options available afterwards. Clear channels of communication are encouraged, further pathways are discussed if there is a confirmatory abnormal chromosomal result. These results are initially relayed by telephone by the specialist midwife informing of the result, allowing the woman to process the information and ask questions. This is very individual and staff are experienced and are sensitive when discussing results allowing time to process and understand the results that have been explained.

The woman is offered an appointment to attend a face to face consultation within a time scale that the woman feels comfortable with. Women are signposted at the face to face consultation to ARC, DSA, SOFTUK. There is also the opportunity to arrange for the woman/ partner to discuss with parents who have children with Down syndrome if this may be helpful (again this is an individual basis).

Alternatively other pathway options can be considered for the ongoing management of pregnancy including referral to Genetics. The objective being that women and their partner are given as much information as they require to make an informed decision and we as health care professionals would support them in this process.

www.arc-uk.org

enquiries@soft.org.uk

www.downs-syndrome.org.uk