Sgrinio Cyn Geni Cymru
Antenatal Screening Wales

Screening Handbook for Midwives

5th Edition
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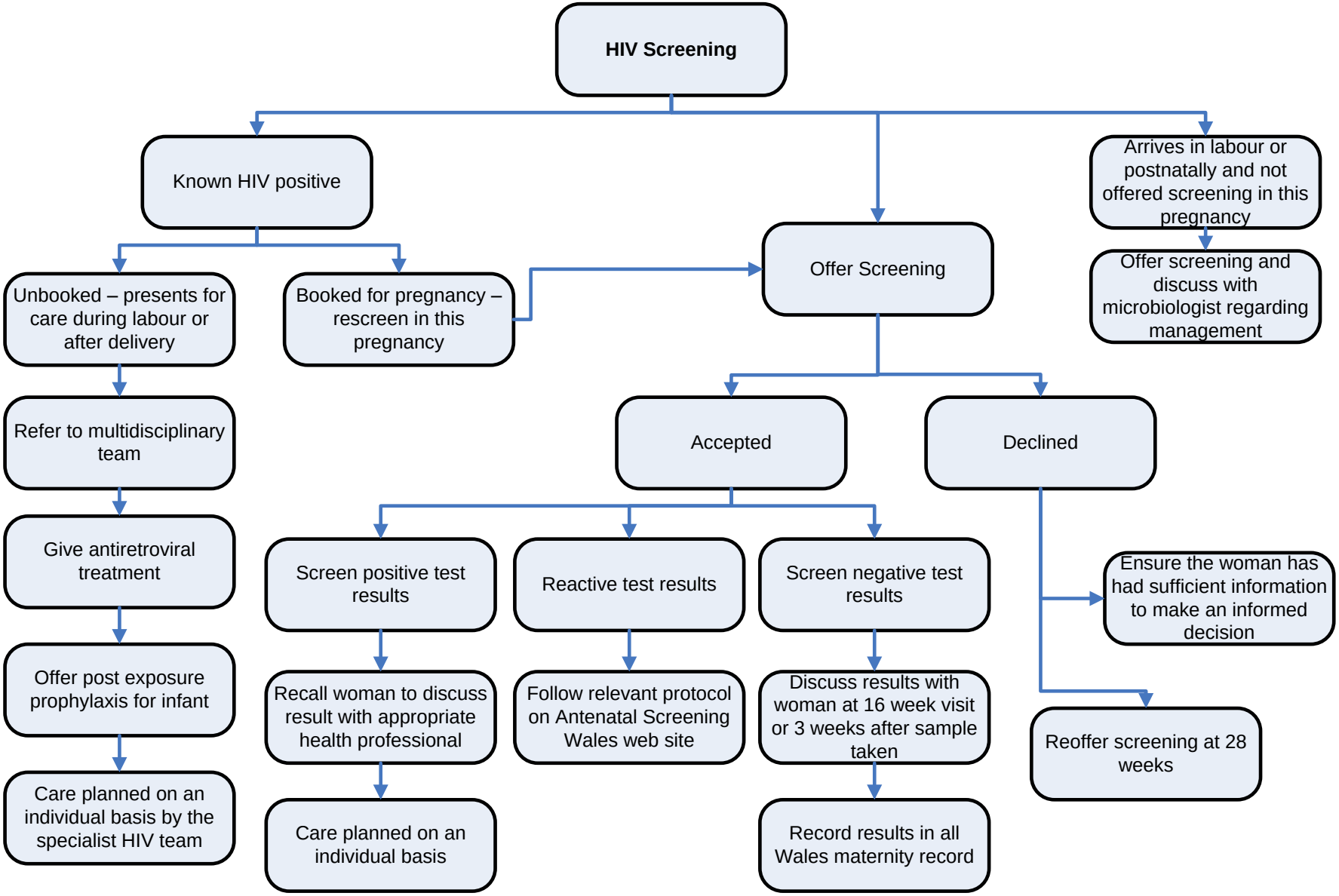
GIG CYMRU NHS WALES
Iechyd Cyhoeddus Cymru
Public Health Wales

Contents

HIV Screening Pathway



HIV (human immunodeficiency virus) Clinical Information

Aim

Antenatal screening for HIV is to identify women who have an established HIV infection so that treatment and care can be offered to reduce the risk of mother to baby transmission of the virus. The identification and treatment of HIV also has considerable health benefits for the woman.

Clinical Information

HIV is a retrovirus that attacks and destroys T-lymphocytes, resulting in immune-suppression that eventually leads to acquired immune deficiency syndrome (AIDS). Vertical transmission of the virus from mother to fetus or baby can occur during pregnancy, at delivery or postnatally through breastfeeding. Two forms of the virus have been identified: HIV-1 and HIV-2. The commonest and most virulent form is HIV-1.

HIV Infection and Pregnancy

- Infants and young children who acquire HIV have an exceptionally high risk of morbidity and mortality, and half will die before their second birthday if they do not receive treatment.
- Without intervention mother to child transmission (MTCT) is 15-25%. Intrauterine infection is extremely rare but the baby can be infected during the birth process.
- With correct treatment the risk of MTCT can be less than 0.2%.

Global Incidence and Prevalence

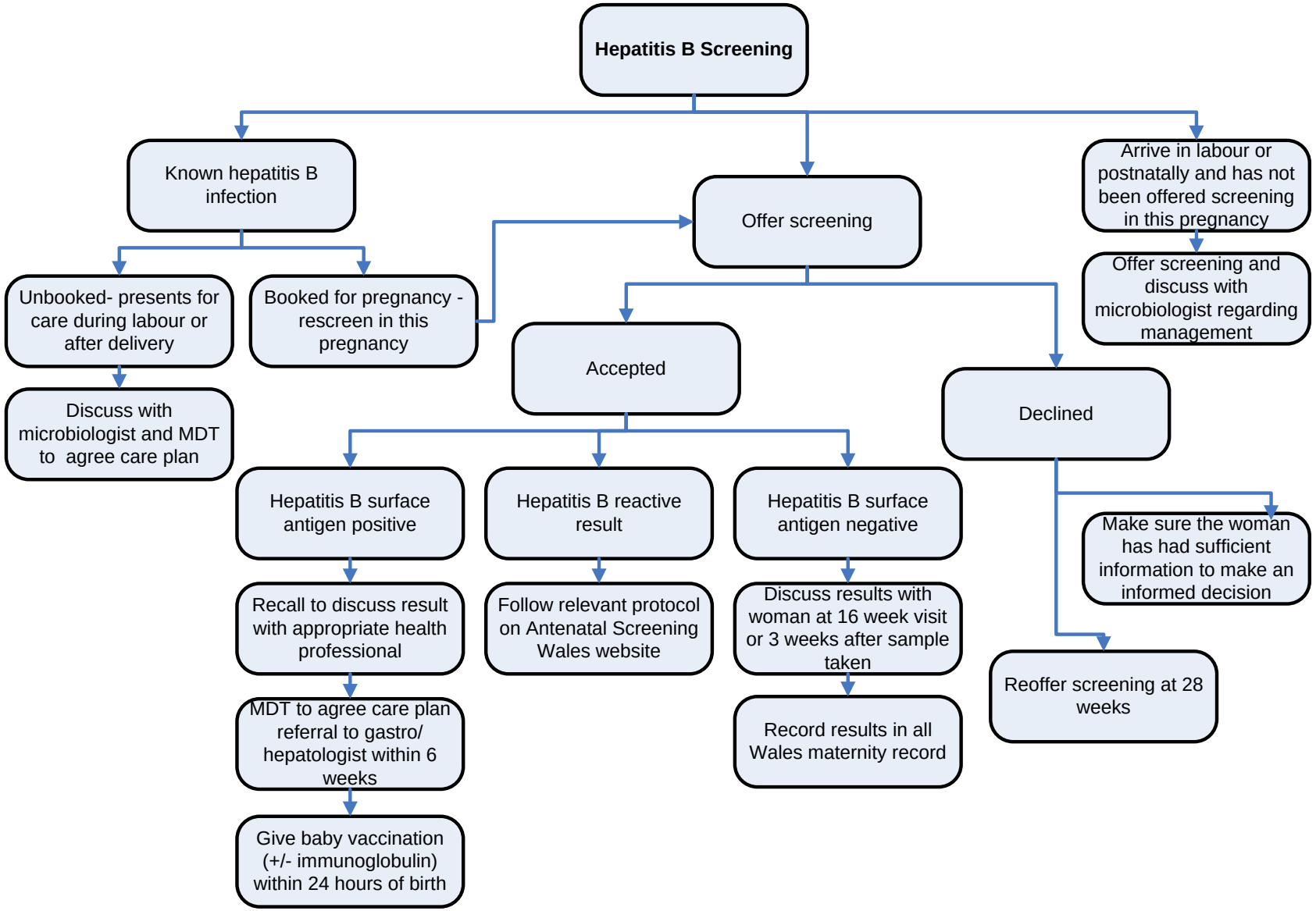
- At the end of 2015 around 36.7 million people globally were living with HIV.
- In 2015 – 2.1 million people became newly infected with HIV.
- In 2015 – 1.1 million people died from AIDS-related illnesses.
- By the end of 2015 there were17 million people accessing antiretroviral therapy.
- Over 90% of infected babies live in Sub-Saharan Africa.

UK/ Wales

- In 2014 HIV prevalence in pregnancy was 1.5 per 1000 women.
- In the UK and Ireland most pregnant women living with HIV are now already diagnosed by the time of conception. The proportion of affected women who are on antiretroviral therapy at conception is currently 60%.
- Effective interventions mean an increasing proportion of women are able to have a vaginal delivery (over 40% by 2014).
- Mother to child transmission of HIV was diagnosed in 110 children born in the UK between 2006 and 2013 (65 were born to mothers undiagnosed during pregnancy).

Between 2012 and 2014 there were just 7 mother-to-child transmissions among nearly 3,300 babies born to diagnosed women living with HIV, corresponding to an MTCT rate of 0.27%.

Hepatitis B Screening Pathway



Hepatitis B Clinical Information

Aim

Antenatal screening for hepatitis B is to enable the identification of women who are infected with hepatitis B whose infants will be at significant risk of contracting hepatitis B at or around the time of delivery. This will enable the offer of post-exposure prophylaxis to the neonate.

Hepatitis B is an infectious disease of the liver caused by the hepatitis B virus and can be detected in blood, saliva and semen and transmitted:

- vertically from mother to baby
- through contact with contaminated blood
- through sexual contact.

Possible Outcome from a Hepatitis B Infection

- Recovery and immunity.
- Persistently infected or chronic carrier state. Between ten and fifty percent of chronic carriers will develop cirrhosis leading to premature death in approximately 50%.
- Fulminant hepatitis (less than1% of symptomatic cases).

Neonatal Implications

- Vertical transmission (mother to infant) of infection occurs in ninety percent of pregnancies where the mother is hepatitis B e antigen positive and in about ten percent of surface antigen positive, e antigen negative mothers.
- Most (>90%) of infected infants become chronic carriers.
- The risk of vertical transmission can be reduced by ninety percent by vaccinating the infant appropriately.

Prevalence

Hepatitis B is endemic worldwide, apart from isolated communities, with very high carriage rates (up to 20%) particularly in South and East Asia, but also in Southern Europe, Central and South America, Africa and Eastern Europe.

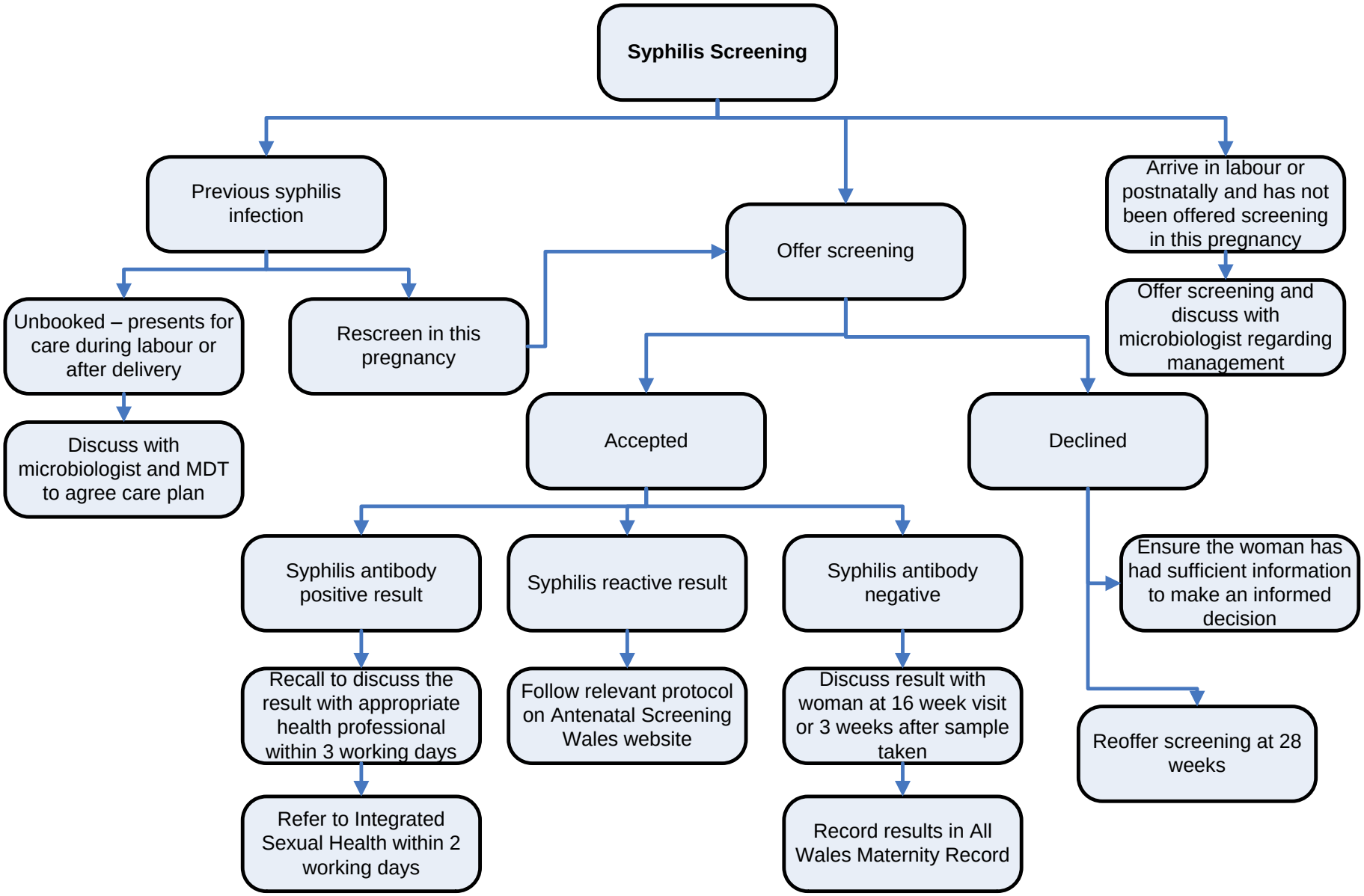
Worldwide, there are an estimated 240 million chronically infected persons.

UK/ Wales Incidence in Pregnant Women

This varies across ethnic groups and is higher from women born in countries where disease is endemic (e.g. 67% of carriers are women born in Africa, China or South Asia) but overall the incidence of hepatitis B infection in pregnant women in the UK is 0.15% (1 - 2 per 1000 women).

There were 66 pregnant women who were hepatitis B positive in Wales in 2015.

Syphilis Screening Pathway



Syphilis Clinical Information

Aim

Antenatal screening for syphilis is to identify women who have syphilis in early pregnancy and offer appropriate treatment to substantially reduce the risks of the fetus contracting congenital syphilis. The identification and treatment of this communicable disease also has potential health benefits for the mother.

Syphilis results from infection by the spirochete bacterium, treponema pallidum. Humans are the only host, and transmission can occur through sexual contact (adult syphilis) or following transmission across the placenta during pregnancy from an infected mother to her fetus (congenital syphilis).

Syphilis is passed from person to person through direct contact with a syphilis sore (chancre). Sores occur mainly on the external genitals, vagina, anus or in the rectum. They can also occur on the lips or in the mouth.

Stages and Symptoms of Maternal Infection and Chance of Transmission to Fetus

Stage of Infection	Maternal Symptoms	Risk of Vertical Transmission to Fetus in Untreated Mother
Primary syphilis	sore (chancre)	70% - 100%
Secondary syphilis	include disseminated disease including fever, malaise, maculopapular rash, hepatitis, meningitis, renal damage	Similar to primary syphilis
Latent syphilis	asymptomatic but moderately infectious	10% - 40%
Tertiary syphilis	2 to 40 years after infection many major symptoms including cardiovascular syphilis, neurosyphilis	Rare

Congenital Syphilis

Syphilis can be transmitted across the placenta at any stage of pregnancy and if untreated is associated with prematurity, low birth weight, non-immune hydrops and intrauterine death:

- Over 60% of fetus's with an infectious mother will be affected.

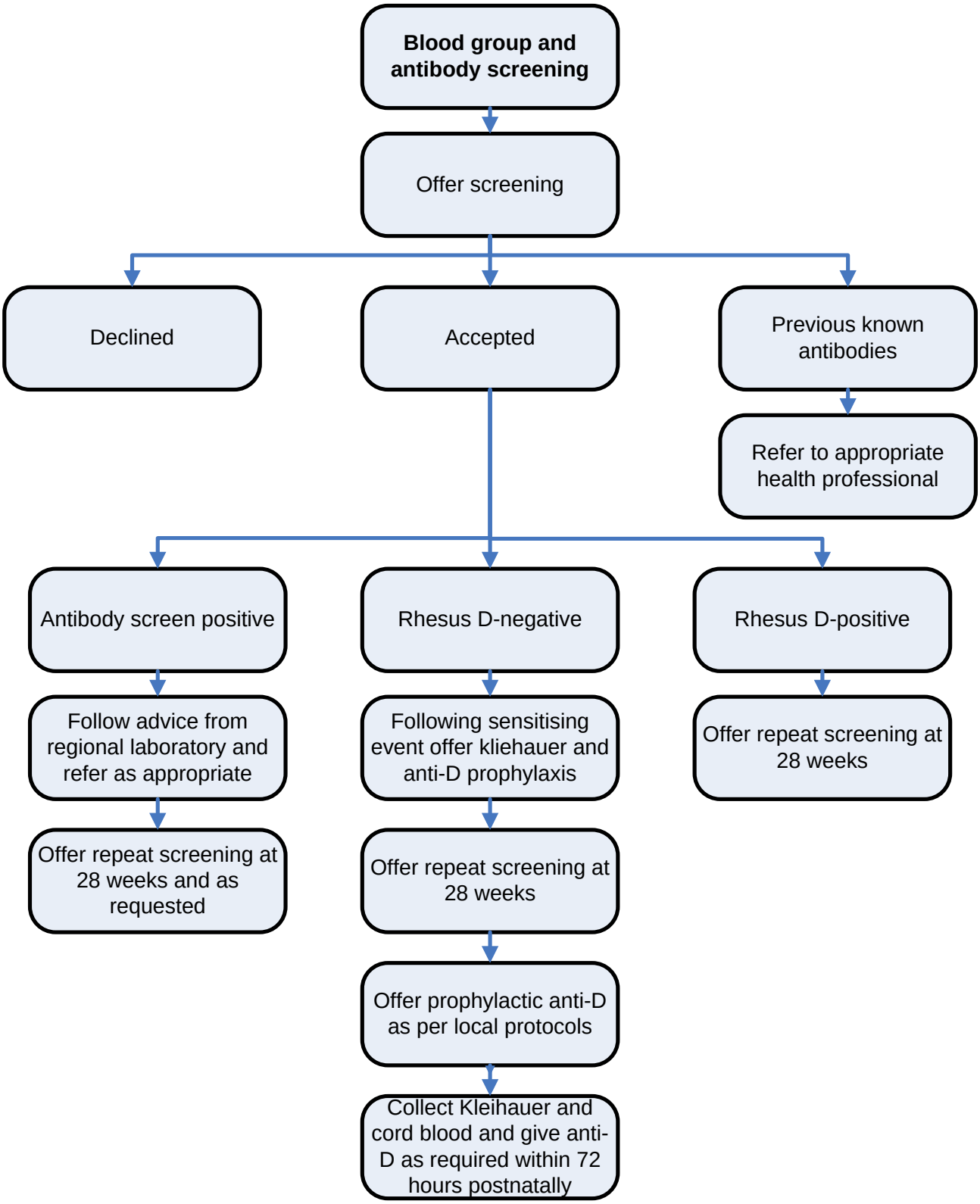
Incidence

- The total number of cases of infectious syphilis reported in Wales in 2014-2015 was 43, a 59% increase from the previous year.
- The proportion of cases acquired heterosexually has varied from 19% in 2008, to 44% in 2012-2013 and 40% in 2014-2015.
- More than 90% of the new cases of syphilis between 2012 and 2015 were in men.

Screening Test Result

- A negative syphilis screening test result means the woman does not have syphilis infection at time of testing.
- Syphilis screening tests cannot always distinguish between syphilis and other non communicable diseases, e.g. yaws, pinta, bejel or a previously treated syphilis infection. The laboratory result therefore needs expert interpretation by a consultant microbiologist/ virologist before the result is issued.

Blood Group and Antibody Screening Pathway



Blood Group and Antibodies Clinical Information

Aim

Antenatal screening for blood group and antibodies should be offered to all pregnant women in early pregnancy, irrespective of previous screening results as an integrated part of their antenatal care. If any antibodies are found, particularly anti D, anti Kell, or anti c, these may indicate a risk of haemolytic disease of the fetus and newborn (HDFN) and the antibodies can be monitored and appropriate obstetric management advised.

There are four main blood groups: group O, group A, group B and group AB. There is also another blood factor called the Rhesus (Rh) group and people have a blood group and Rh group, e.g. group O Rhesus D positive. RhD factor is the protein found in red cells in about 85% of people and its presence denotes a person is RhD positive and absence denotes the person is RhD negative.

Where the woman is RhD negative and the baby is RhD positive there is a possibility of maternal antibodies being produced (alloimmunisation) and passing from the maternal bloodstream into the fetus causing HDFN. Rhesus negative pregnant women should be offered prophylactic anti-D where there is a risk of alloimmunisation following a sensitising event and as part of normal antenatal care in the third trimester.

Inheritance Patterns

In genetic terms, the RhD positive allele is dominant (D) and the RhD negative allele (d) is recessive. Consequently, there are three possible genetic pairs for Rh alleles.

Genes	Blood Type
DD	RhD positive
Dd	RhD positive
dd	RhD negative

There are a number of possible combinations of RhD types in parents but the possibilities outlined here are only those that occur where the woman is RhD negative, because RhD positive women are not affected by this issue.

- If both of the parents are RhD negative, all babies will be RhD negative.
- If the woman is RhD negative and the biological father is RhD positive, the genetic (and potential clinical) outcomes are dependent upon whether the baby's biological father is homozygous RhD positive or heterozygous RhD positive.
- If the father is homozygous RhD positive (DD), all of his children will inherit one RhD positive allele from him (and one RhD negative allele from their mother) and all of the couple's babies will be heterozygous RhD positive.
- If the father is heterozygous RhD positive (Dd), his children will have a 50% chance of inheriting an RhD positive allele from him and a 50% chance of inheriting an RhD negative allele from him. (Around 55% of RhD positive men are thought to be heterozygous).
- If the baby inherits the RhD positive allele from their father, they will be heterozygous RhD positive.
- If the baby inherits the RhD negative allele from their father and mother they will be RhD negative.

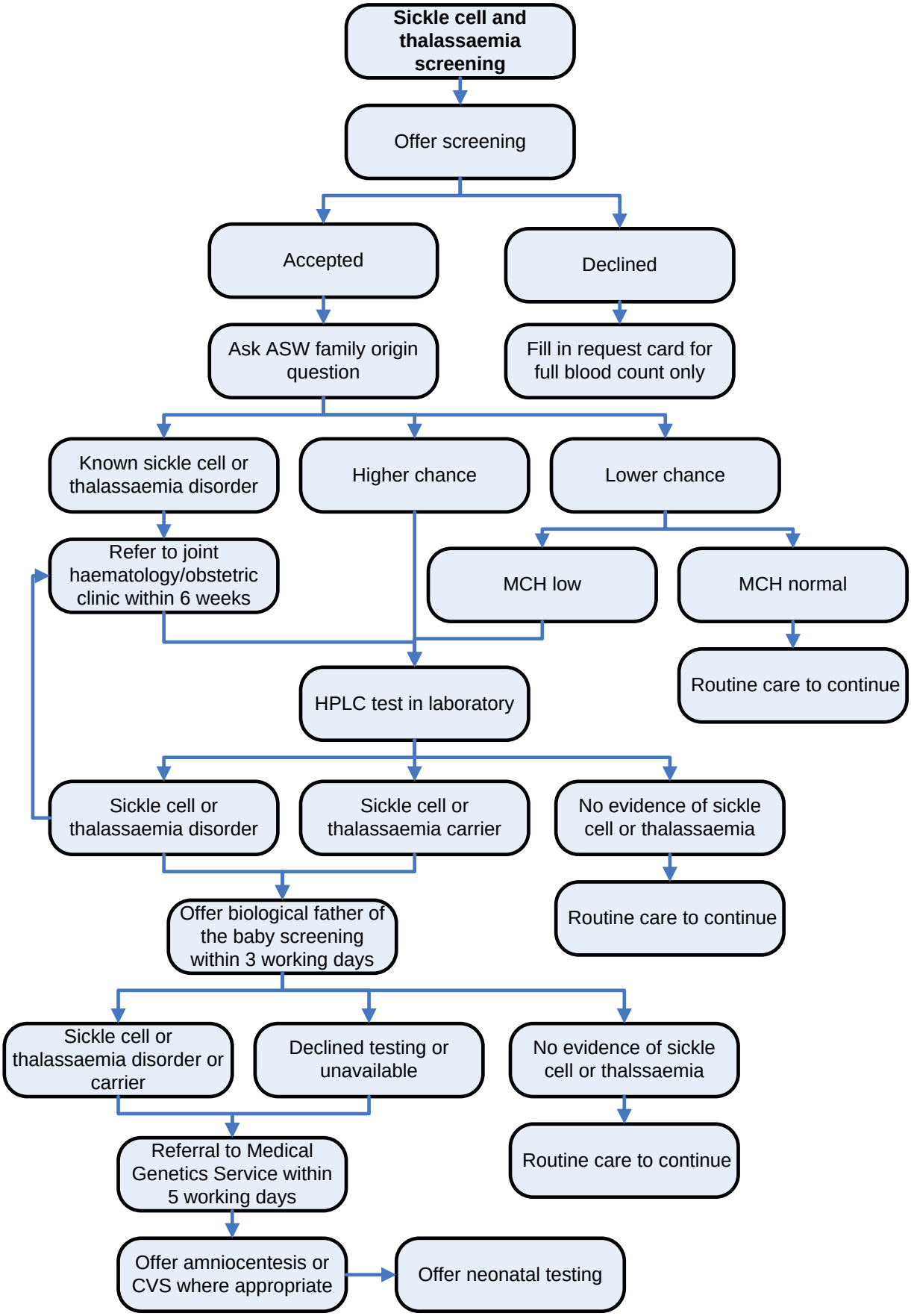
		Paternal	
		D	D
Maternal	d	Dd	Dd
Maternal	d	Dd	Dd

		Paternal	
		D	d
Maternal	d	Dd	dd
Maternal	d	Dd	dd

Incidence

The rate of alloimmunisation in the UK ranges between 0.17% and 0.28% and mortality caused by HDFN is 1.6/ 100 000 births.

Sickle Cell and Thalassaemia Screening Pathway



Sickle cell and Thalassaemia Clinical Information

Aim

Antenatal screening for sickle cell and thalassaemia is to identify women who have a high chance of having a fetus affected by a sickle cell disorder or thalassaemia major to enable decisions about whether to have invasive testing and continuing the pregnancy.

Sickle cell disorders

These are genetic conditions where an individual inherits sickle haemoglobin which affects the ability of the haemoglobin to function normally and results in chronic multi-system organ disease. The characteristics of sickle cell disease are:

- chronic haemolytic anaemia
- jaundice
- painful crisis
- organ damage where ‘sickling’ occurs
- susceptibility to infections
- strokes in childhood.

Thalassaemia

These are haemoglobin gene variants which affect the production of globin chains. They are classified according to the chain which is inefficiently produced, i.e. alpha or beta thalassaemia.

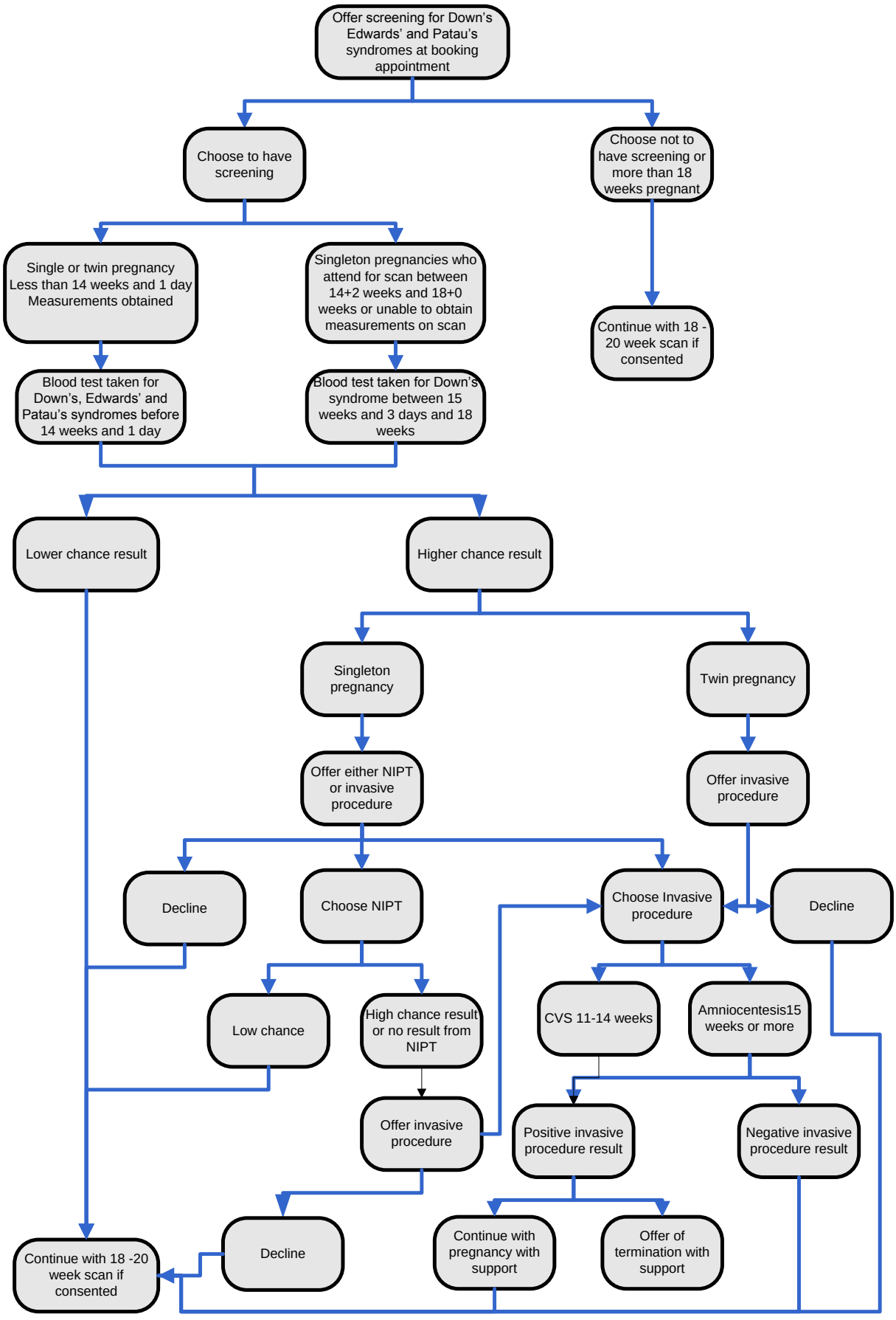
- Alpha thalassaemia is an inability to produce alpha globin chains. The fewer alpha globin genes the more serious the condition. Alpha thalassaemia major is incompatible with extra uterine life.
- Beta thalassaemia is an absence or reduced output of the beta globin chain synthesis and result in a reduced production of haemoglobin causing varying degrees of anaemia. Beta thalassaemia major can be life threatening requiring regular blood transfusion and iron chelation for survival. Beta thalassaemia major without treatment usually causes severe anaemia between the age of three and eighteen months and typically children do not live beyond early childhood.

Prevalence

Wales has a low prevalence of sickle cell disorders and thalassaemia major with approximately 10 pregnancies a year where due to the mother and father’s result the baby was predicted to be 'at risk' of having either sickle cell disorder or thalassaemia major.

Pathway for Screening for Down's, Edwards' and Patau's Syndromes

Note: At all stages in the pathway women should be offered appropriate counselling and support



Down's, Edwards and Patau's Syndromes Clinical Information

Aim

Antenatal screening for Down's, Edwards' and Patau's syndromes offers women the choice to identify whether they have an increased chance of having a baby affected by one of these syndromes to enable them to decide whether to have further testing and, if necessary, make choices about continuing the pregnancy.

The screening involves:

- the offer of screening in the first trimester for Down's, Edwards' and Patau's syndromes using the combined test in singleton or twin pregnancies.
- If too late for this test, or unable to get the required ultrasound measurements, the offer of a quadruple test for Down's syndrome for singleton pregnancies only.

For woman with a higher chance result from the combined or quadruple test:

- The offer of an invasive test to singleton and twin pregnancies.
- Or the choice of non-invasive prenatal testing (NIPT) **to women with a singleton pregnancy only**.

Predisposing factors for Down's, Edwards' and Patau's syndromes are: increasing maternal age; and a family history of one of the syndromes which indicates an increased risk for either Down's, Edwards' or Patau's syndrome.

People with Down's syndrome are affected in different ways, all have some learning disability, most can lead nearly independent adult lives but some need more support than others.

Most babies with Edwards' or Patau's syndrome will die before they are born or shortly after birth. Of the babies who survive, about 1 out of 7 or 8 (12-13%) will live for more than a year. People with Edwards' syndrome or Patau's syndrome will have lifelong learning disabilities.

Characteristics of Down's syndrome are variable but include:

- their birth weight is lower than average and they may put on weight at a slower rate than other babies.
- they may have looser muscles and joints than other babies.
- they often have eyes that slant upwards and outwards. Their eyelids can have an extra fold of skin (known as the epicanthic fold) which appears to make the slant more noticeable (almond-shaped eyes).
- the back of the baby's head may be flatter than average.
- many babies with Down's syndrome have a single crease which runs right across the palm of their hands.

Associated Health Problems of Down's syndrome

- about 1 out of 2 (50%) children with Down's syndrome will have a heart problem, and around 1 out of 4 (25%) may need an operation. Some of the heart problems are life-threatening.
- visual problems are much more likely, or more likely to happen at a younger age, in people with Down's syndrome. Congenital cataracts are 10 times more likely and there is also a risk of

infantile glaucoma. Around 6 out of 10 (60%) children with Down's syndrome will have some hearing loss which may cause problems with speech and language.

- around 1 out of 14 (7%) children with Down's syndrome have problems with their digestive tract, which in some cases may need treatment and surgery.
- about 1 out of 7 (15%) children with Down's syndrome will die before the age of five.
- infections of the ears, nose and throat are more likely.
- Leukaemia (blood cancer) is more common.
- most children with Down's syndrome will learn to walk and talk. Around 8 out of 10 (80%) will go to mainstream primary school and learn to read and write
- more and more adults with Down's syndrome are living more independently and are included in their community. Many adults are capable of work and may live in their own accommodation, with support. Some need more support than others and there is no way of knowing when the baby is born how much support they might need in adulthood.
- with good healthcare, people with Down's syndrome can live into their 60s.
- Alzheimer's disease (a form of dementia) may affect people with Down's syndrome at an earlier age than other people. Symptoms of Alzheimer's disease, such as memory loss, happen in about 3 out of 4 (75%) people with Down's syndrome who are aged over 60.
- with appropriate medical care, most children and adults with Down's syndrome can lead healthy and fulfilled lives.

Incidence of Down's syndrome

In Wales, Down's syndrome occurs once in every 415 pregnancies with the incidence increasing with increasing maternal age.

Characteristics of Edwards' and Patau's syndromes include:

- small, abnormal-shaped head
- low-set ears
- small jaw and mouth

Associated Health Problems of Edwards' and Patau's syndromes

Babies with Edwards' syndrome will have a narrow but often serious range of problems.

- Significant developmental delay (all babies).
- Heart problems (many babies).
- Feeding difficulties (many babies).
- Medically fragile, especially with respiratory problems (most babies, especially when small).
- Cleft lip and palate (some babies).
- Other problems, including being more prone to conditions such as urinary tract infections (UTIs).

At the other end of the scale, research shows, for example, that:

- babies and children make progress, however slowly
- older babies and children show some level of communication
- some will stand and walk with help, and
- parents consistently report a high quality of life for their babies and children, because they are involved in family activities.

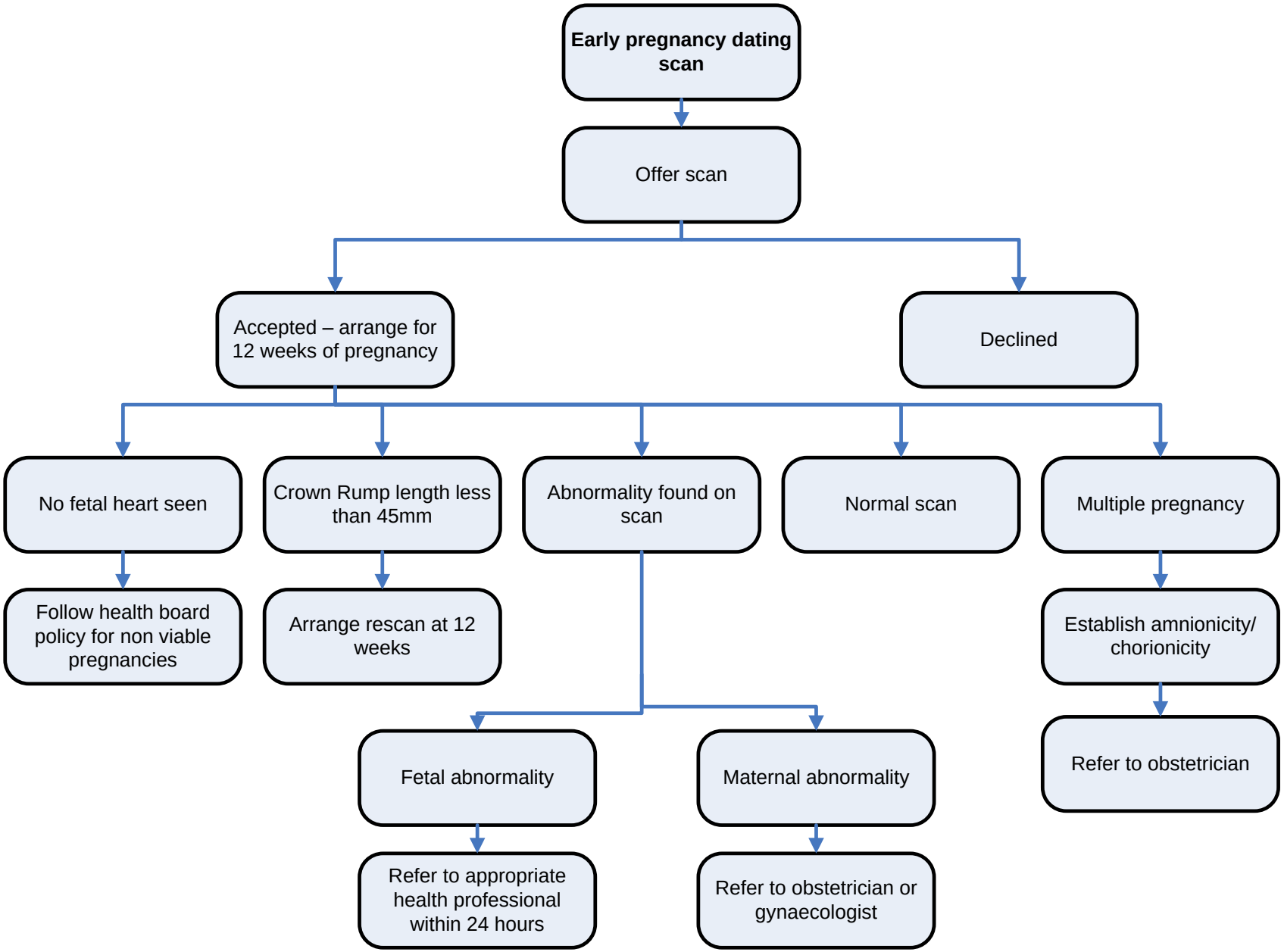
Incidence of Edwards' and Patau's syndrome

In Wales, Edwards' syndrome occurs once in every 1656 pregnancies and Patau's syndrome occurs once in every 4201 pregnancies with the incidence of both syndrome increasing with increasing maternal age.

Results

Between 97% and 98% of women who choose to have screening will receive a lower chance Down's, Edwards' and Patau's syndrome screening result. Around 2% to 3% of women will receive a higher chance result (result between 1 in 5 and 1 in 150) and will be offered either non-invasive prenatal testing (NIPT) or an invasive test.

Early Pregnancy Ultrasound Scan Pathway



Early Pregnancy Ultrasound Scan Clinical Information

Aim

The early pregnancy ultrasound scan is offered to determine viability, the gestational age and to detect multiple pregnancies (fetal number and chorionicity/ amnionicity). Some major fetal anomalies may be detected, but this is not the primary purpose of this scan. Measurements to determine the gestational age are required for the Down's, Edwards' and Patau's syndromes screening programme and also an additional measurement if the scan is before 14 weeks and 1 day of pregnancy (maximum CRL 84.0mm). Using ultrasound derived gestation reduces the need for post term induction of labour. Where first trimester screening for Down's, Edwards' and Patau's syndromes is provided, the woman will receive an earlier screening test result.

Information Obtained from the Early Pregnancy Scan

- Can identify a non viable pregnancy, a fetal demise or an empty sac.
- Will confirm if the pregnancy is intra-uterine.
- May identify multiple pregnancies.
- Measurements obtained are used to calculate the correct gestation and provide an accurate estimated date of delivery (EDD).
- Provides the measurements for Down's, Edwards' and Patau's syndromes screening:

  Combined test

  - Crown Rump Length (CRL) 45.0mm to 84.0mm and
  - Nuchal Translucency (NT)

  Quadruple test

  - Head Circumference (HC) 98mm to 147mm.

Possible Abnormalities Detected

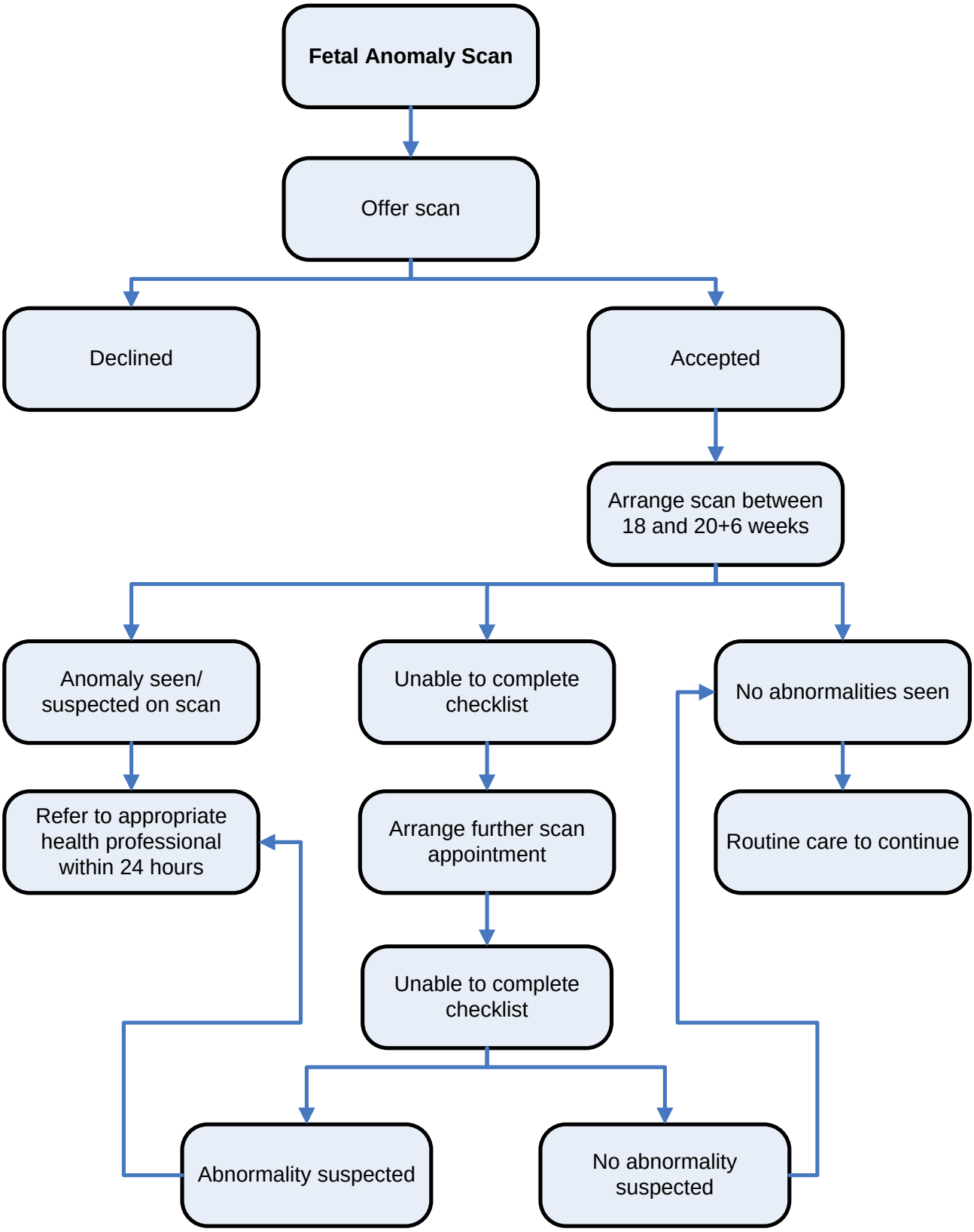
Some major abnormalities may be detected:

- Anencephaly
- Enlarged Nuchal Translucency (over 3.5mm)
- Omphalocele
- Renal agenesis

Limitations of the Early Pregnancy Scan

- Visualisation of the fetus will affect what can be seen on the scan. Some things that affect visualisation are:
  - Woman's body mass index
  - Position the fetus is lying
  - Uterine fibroids
  - Abdominal scarring
- Scans may give false reassurance as:
  - Some abnormalities may not become detectable until later in the pregnancy
  - Absence of an abnormality does not guarantee a normal baby as many conditions cannot be diagnosed by ultrasound scans
  - Fetuses with chromosomal abnormalities cannot always be diagnosed by ultrasound.
- Detection rates of abnormalities depend on the type of anomaly

Fetal Anomaly Ultrasound Scan Pathway



Fetal Anomaly Ultrasound Scan Clinical Information

Aim

The purpose of the fetal anomaly ultrasound scan is to detect significant structural fetal anomalies that are likely to have an adverse effect on the health of the mother and/ or baby and for which an effective intervention is available and warranted at 18 weeks and 0 days to 20 weeks and 6 days of pregnancy.

Information Obtained from the Anomaly Scan

At the anomaly scan the structures that are identified on the ASW standard checklist must all be visualised for the scan to be completed. The woman will be offered one further scan if not all structures are identified. If the check list **is not completed at the second visit** the woman **will not be rescanned** and this will need to be documented in the woman's notes.

Limitations of the Anomaly Scan

- Visualisation of the fetus will affect what can be seen on the scan. Some things that affect visualisation are:
    - Woman's body mass index
    - Position the fetus is lying
    - Uterine fibroids
    - Abdominal scarring
- Scan may give false reassurance as:
    - Some abnormalities may not become detectable until the third trimester
    - Fetuses with chromosomal abnormalities cannot always be diagnosed by ultrasound.
    - Detection rates of abnormalities depend on the type of anomaly as many conditions cannot be diagnosed by ultrasound scans see chart below:

The problem	The chance of the problem being seen on an ultrasound anomaly scan at 18 to 20 weeks
Spina bifida (skin or bone not covering the spinal cord) Spina bifida is a fault in the development of the spine and spinal cord which leaves a gap in the spine. The spinal cord connects all parts of the body to the brain.	**90%**
Major heart condition, for example: **tetralogy of fallot** Tetralogy of fallot is a serious heart condition where the heart has not developed in the same way as a normal heart in the womb. This condition will need surgery usually in the first year of birth.	**40%**
Autism Autism cannot be picked up on scan as there is no structural abnormality. Autism is a lifelong developmental disability that affects how a person communicates with, and relates to, other people. It also affects how they make sense of the world around them.	**0%**