Familial Hypercholesterolaemia

What is the disease?

- FH is due to mutations of our DNA which prevent our bodies breaking down cholesterol
- Patients will have very high cholesterol from birth
- Very high cholesterol increases our chances of having heart disease such as a heart attack

Why screen for this disease?

- This disease is thought to affect 1 in 500 people meaning 110,000 in the UK will be affected
- BUT only 18,000 people have been diagnosed. Where are the rest?
- If you have FH there is a 50% chance that you'll pass this on to to your children
- If picked up early FH is easily controlled and can save the NHS money as it prevents more costly treatment later

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Who gets screened?

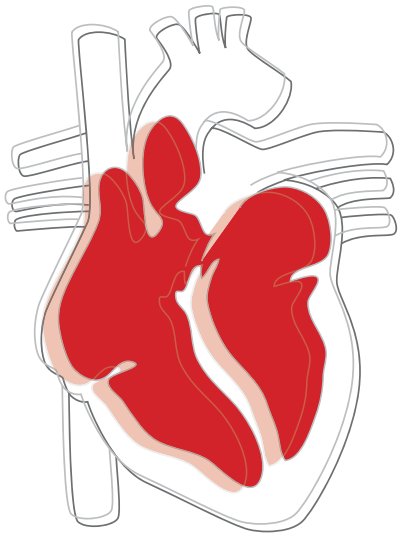
- Adults who have very high cholesterol
- As the disease is inherited once an adult is found to have the disease other family members will be screened too
- Genetic testing is used to confirm disease in a patient

What are the advantages of identifying this disease?

- Person at less risk of having heart problems later on in life
- Cheaper for the NHS as saves money on costly procedures later
- Gives patient motivation to look after themselves better, i.e. eat healthier and exercise more

Are there any disadvantages?

- Could the doctor get it wrong?
- If disease only causes problems later on in life do patients need to take treatment now?
- Will a patient be able to get life insurance if they have this disease? If not, they may not want to have the test



Down's Syndrome

What is the disease?

- The disease results from a chromosomal defect in which there's an extra chromosome 21. In normal cells there are 46 chromosomes however those with DS have 47 chromosomes
- Common traits looked for at birth include: low muscle tone and reduced strength, flat facial profile, upward slant to the eyes
- DS children will have learning disabilities but the degree of this disability can vary
- Increased risk of congenital heart defect, infections, respiratory problems, obstructive digestive tracts, childhood Leukaemia

Why screen for this disease?

- To give a choice for the parents
- Early involvement can lessen the degree of learning disability

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Who gets screened?

- Every woman is given access to DS screening
- Incidence of this disease increases with age resulting in more mature women opting for the screen
- Parents with chromosomal defects are at higher risk
- If parents already have one DS child then there's an increase in risk by 1% that their next chid will have DS
- Biochemical tests which measure the levels of hormones in the blood and ultrasounds which measure fluid in the foetus are carried out initially
- Follow up genetic tests can be carried out:

    Amniocentesis takes a sample of the amniotic fluid that surrounds the foetus to be analysed

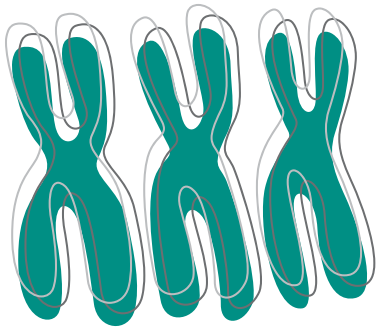
    Chorionic Villus Sampling takes a small piece of the placenta to be analysed

What are the advantages of identifying this disease?

- Early intervention in a DS child's development can lower the degree of learning disability
- Parents can plan ahead

Are there any disadvantages?

- The screening test amniocentesis can cause miscarriage usually within 3 weeks of the test.
- Screening tests are not conclusive: false positives vary depending on the screening procedure.



Breast Cancer

What is the disease?

- Most common cancer in the UK - 46,000 people are diagnosed every year
- Female to male incidence is 100:1
- Many factors are implicated in the cause

Why screen for this disease?

- It is very common and small tumours are difficult to detect by self-examination
- It is very cost effective
- It is important to identify disease before it spreads. Evidence shows that regular breast screening reduces deaths by 35% but there are still 10, 200 female deaths per year in UK
- Breast screening can also detect other diseases which may develop into cancer over time, however it is not known what disease will develop into cancer until the cancer is found

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Who gets screened in England?

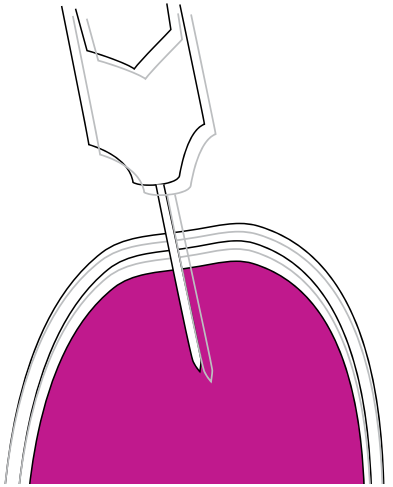
- All females aged over 50 and registered with a GP every three years
- Life time risk increases with age. Breast cancer is less common before menopause
- Male breast cancer is uncommon and the test is difficult to undertake in males
- Screening is performed by a mammography, which is a low dose of X-rays

What are the advantages of identifying this disease?

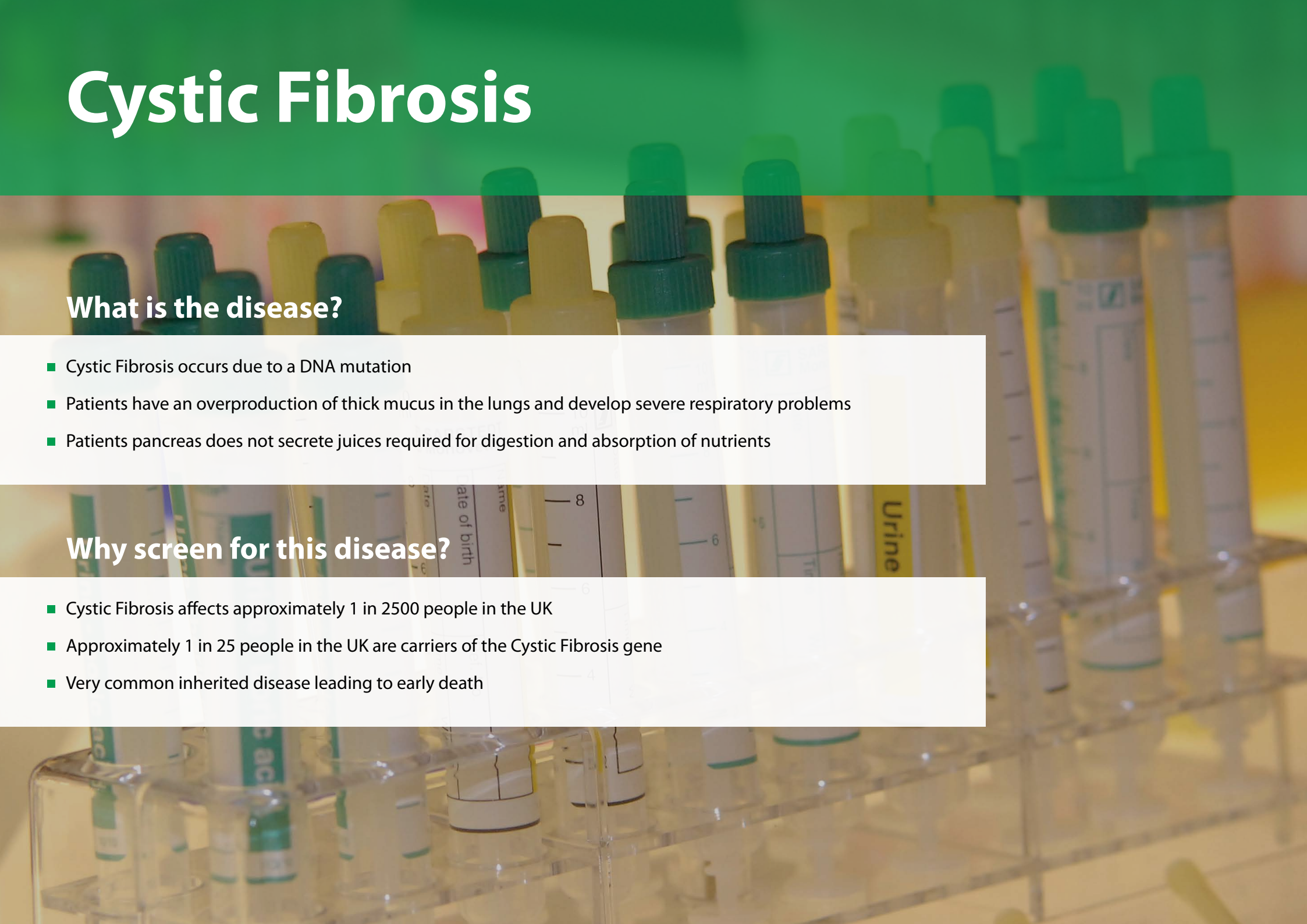
- If breast cancer is identified early it can be cured
- Lower psychological impact as the surgery is less severe when the disease is in the early stages

Are there any disadvantages?

- A mammography is often uncomfortable and sometimes painful
- Like all screening tests, it is not 100% sensitive and the disease may be missed
- It is a sensitive issue for females and can lead to surgical disfigurement



Cystic Fibrosis



What is the disease?

- Cystic Fibrosis occurs due to a DNA mutation
- Patients have an overproduction of thick mucus in the lungs and develop severe respiratory problems
- Patients pancreas does not secrete juices required for digestion and absorption of nutrients

Why screen for this disease?

- Cystic Fibrosis affects approximately 1 in 2500 people in the UK
- Approximately 1 in 25 people in the UK are carriers of the Cystic Fibrosis gene
- Very common inherited disease leading to early death

Who gets screened?

- Every single baby born in the UK since 2007 (US and many other countries have also introduced the screening)
- If disease is not diagnosed at birth, but symptoms appear later, screening is performed again (usually all are diagnosed within first few years of life)
- Genetic testing is performed to confirm presence of Cystic Fibrosis

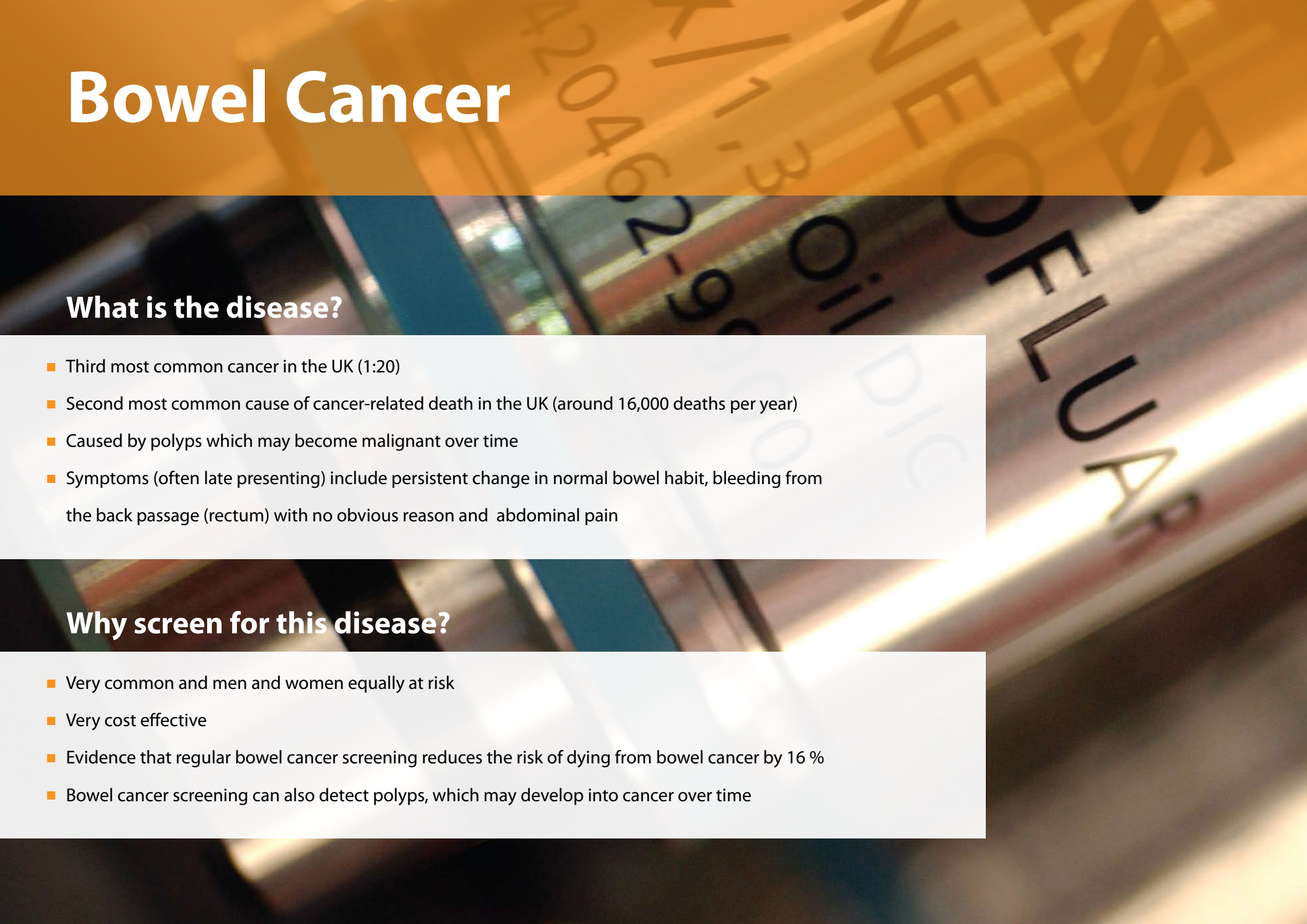
What are the advantages of identifying this disease?

- Treatment can be started straight away, before serious symptoms develop
- Earlier diagnosis and treatment extends life expectancy
- Allows parents to make an informed choice about future pregnancies

Are there any disadvantages?

- There is no cure for Cystic Fibrosis – treatment only marginally extends life expectancy (most patients die by the age of 30)
- Over 1000 mutations are known, only the most thirty mutations are screened, which means many patients could be missed
- Many mutations are very mild, with no symptoms, but patient is still classed as having Cystic Fibrosis

Bowel Cancer



What is the disease?

- Third most common cancer in the UK (1:20)
- Second most common cause of cancer-related death in the UK (around 16,000 deaths per year)
- Caused by polyps which may become malignant over time
- Symptoms (often late presenting) include persistent change in normal bowel habit, bleeding from the back passage (rectum) with no obvious reason and abdominal pain

Why screen for this disease?

- Very common and men and women equally at risk
- Very cost effective
- Evidence that regular bowel cancer screening reduces the risk of dying from bowel cancer by 16 %
- Bowel cancer screening can also detect polyps, which may develop into cancer over time

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Who gets screened in England?

- All men and women 60-69 years registered with a GP, every 2 years until age 69
- People over 70 may may call a helpline
- Risk of developing bowel cancer increases with age: 80% of people diagnosed are around 60 years

How does screening take place?

- Screening by faecal occult blood testing (FOB). Polyps and bowel cancers can bleed and thus blood detected in faeces

What are the advantages of identifying this disease?

- May lead to cancer diagnosis . If bowel cancer detected at an early stage, there is around a 90 % chance of survival
- May also detect diseases, for example ulcerative colitis

Are there any disadvantages?

- FOB tests are quite unpleasant!
- Not 100 % sensitive. There is a chance of cancer being missed if not bleeding at time of sampling
- May lead to cancer diagnosis which has psychological implications
- Not all bowel cancers detected by screening can be successfully treated
- It is a sensitive subject demonstrated in the low uptake of testing – only 52% in the UK are tested