



Newborn Metabolic Screening Program

CUHK-BCM Joint Centre for Medical Genetics
The Chinese University of Hong Kong



Information for Parents
Screening for

Cystic Fibrosis

Enquiries:

(852) 5569 6412 (office hours)

(852) 3505 4219 (voicemail service)

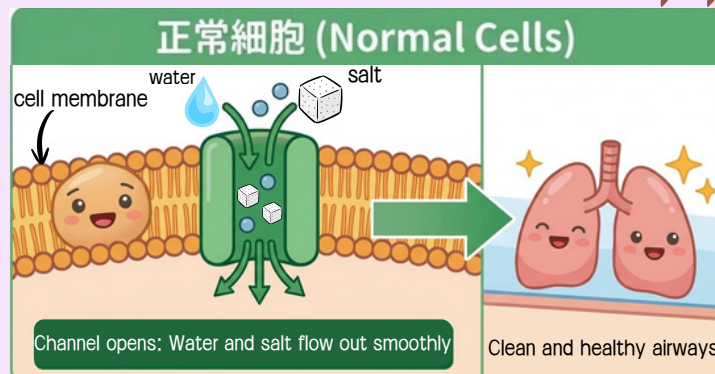
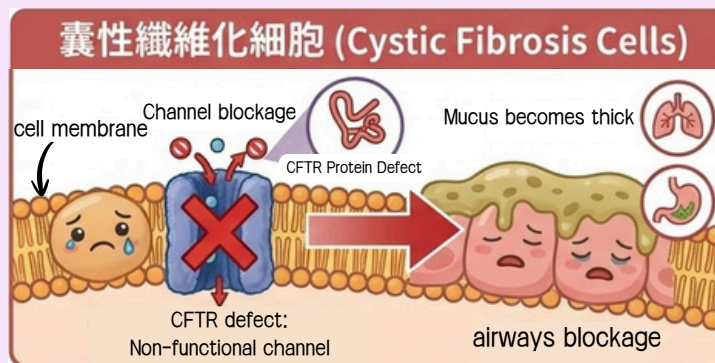
website:



If you wish to join this screening program,
please contact your obstetrician during antenatal period
or contact your paediatrician within 7 days after delivery

What is Cystic Fibrosis?

Cystic fibrosis is an **inherited condition** caused by changes in the CFTR gene, which **affect the function of the CFTR protein**. As a result, thick and sticky mucus can build up in the lungs, pancreas, and other organs, leading to breathing difficulties, digestive problems, and an increased risk of infection.



What are the symptoms of Cystic Fibrosis?

The signs and symptoms of cystic fibrosis can vary from person to person, and some babies may develop symptoms shortly after birth.

Possible features include:

poor weight gain

bulky/greasy stools

meconium ileus

persistent cough

wheezing

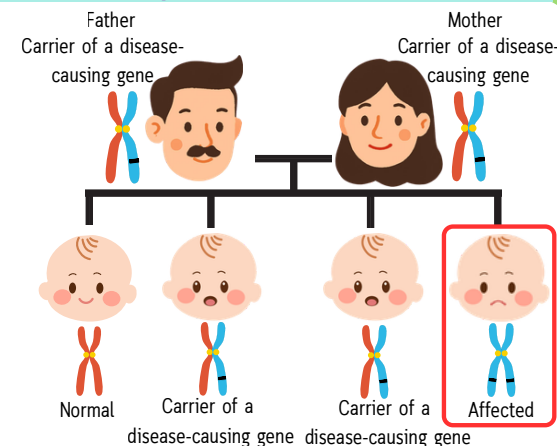
frequent lung infections

salty-tasting skin

digestive problems

Some affected individuals may have an increased risk of chronic lung disease, diabetes, and reproductive problems later in life.

How is Cystic Fibrosis inherited?



Cystic fibrosis is an **autosomal recessive condition**, which means that a baby must inherit one disease-causing CFTR gene variant from each parent to be affected. If both parents are carriers, each pregnancy has 1/4 chance of having Cystic Fibrosis, a 1/2 chance of being a carrier without any symptoms and a 1/4 chance of not carrying any affected gene at all.

Cystic Fibrosis screening and its aim

The Cystic Fibrosis screening aims to detect babies at risk of Cystic Fibrosis as early treatment can prevent severe disability.

A few drops of blood are collected on a card by pricking the baby's heel in its first few days of life, and the concentration of immunoreactive trypsinogen (IRT) is measured. If IRT is elevated, CFTR gene analysis follows. All babies with a positive screening result will undergo further investigations to confirm whether they are affected by Cystic Fibrosis or not.

The incidence of CF varies by race, being relatively rare in Asians.



Outcomes of Cystic Fibrosis patients

Regular visits to the specialist respiratory team are needed for monitoring and management.

Babies with Cystic Fibrosis should follow treatments including airway clearance, enzyme supplements, antibiotics and nutritional support under the instructions of doctors and dietitians.

Doctors and dietitians will provide expert advice to prevent complications from thick mucus accumulation.