

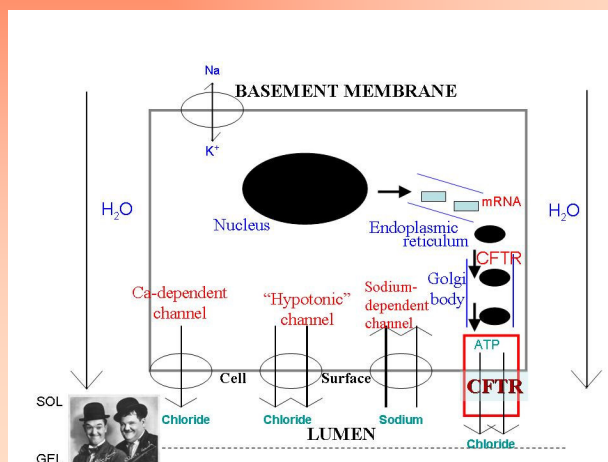
# Newborn Screening for Cystic Fibrosis in South Africa: How & Who?

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## CF & Newborn Screening

### Brief Outline of Cystic Fibrosis



# CF & Newborn Screening

## Cystic Fibrosis in South Africa

Table 1. Frequency of common CFTR mutations in cystic fibrosis patients from different South African population groups

Mutation	White (N = 201)*	Coloured (N = 43)*	Black (N = 14)*
ΔF508	0.76	0.500	-
3272-26A → G	0.040	0.012	-
3120+1G → A	0.005	0.174	0.464
3944delTT	0.037	-	-
G542X	0.017	0.023	-
R553X	0.01	-	-
W1282X	0.01	-	-
N1303K	0.01	-	-
G551D	0.008	0.023	-
R117H	0.0025	-	-
Q493X	0.0025	-	-
S549N	0.0025	-	-
621+1G → T	0.0025	-	-
1717-1G → A	0.0025	-	-
2789+5G → A	0.0025	-	-
3659delC	0.0025	-	-
R1162X	-	0.012	-
G1249E†	-	-	0.036
3196del54†	-	-	0.036
-94G → T†	-	-	0.036
2183delAA†	-	-	0.036
Unknown	0.086	0.256	0.393
Total mutation detection	0.910	0.744	0.607

\* N = number of patients studied.  
† These patients were reported previously.  
- = not observed.

**Carrier Rates**  
**White 1:27**  
**Coloured 1:50**  
**Black 1:14 – 1:59**

# CF & Newborn Screening

## Advantages of Screening

# CF & Newborn Screening

Methods of Screening

**Immuno-reactive trypsinogen (IRT)**

**Molecular genetics (CFTR panel)**

# CF & Newborn Screening

IRT issues in South Africa

**What affects 'normal' levels?**

# CF & Newborn Screening

## CFTR issues in South Africa

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# CF & Newborn Screening

## Follow up issues in South Africa

### Results of first test

Sweat testing

Distance

Counselling / CF Expertise

# CF & Newborn Screening

Where to?

Answer some scientific questions re IRT  
Targetted screening? Equitable?