CARRIER FREQUENCY IN COMMON AUTOSOMAL RECESSIVE GENETIC CONDITIONS

This document is a reference guide to be **used in conjunction with the Clinical Prioritisation Criteria for Genetic Health Queensland**, and lists the carrier frequency in common autosomal recessive genetics conditions.

Condition	Carrier frequency
Alpha thalassaemia	1:30 Southeast Asian, African, and Mediterranean
Alpha-1 antitrypsin deficiency	1:11 Scandinavian; 1:35 European
Ataxia telangiectasia	1:81 Moroccan and Tunisian Jewish; 1:100 European
Autosomal recessive polycystic kidney disease	1:70 All
Beta thalassaemia	1:25 Mediterranean, Middle Eastern, Central Asian, and Southeast Asian
Congenital adrenal hyperplasia	1:35 Middle Eastern; 1:50 European; 1:72 Japanese
Cystic fibrosis	1:25 European
Friedreich Ataxia	1:60 European, Middle Eastern, South Asian, and North African
Haemochromatosis	1:10 European; 1:33 Hispanic; 1:775 African American; 1:1000 Asian
Hearing loss (non-syndromic, GJB2 related)	1:12 East Asian; 1:21 Ashkenazi Jewish; 1:40 Other
Phenylketonuria	1:26 Turkish; 1:33 Irish; 1:50 European; 1:50 East Asian; 1:200 Japanese
Sickle cell disease	1:10 African American
Spinal muscular atrophy	1:45 European; 1:48 Asian; 1:56 Jewish; 1:77 Hispanic, 1:100 Sub-Sahara African
Tay-Sachs disease	1:30 Ashkenazi Jewish; 1:250 Other
Wilson disease	1:40 European; 1:90 Other