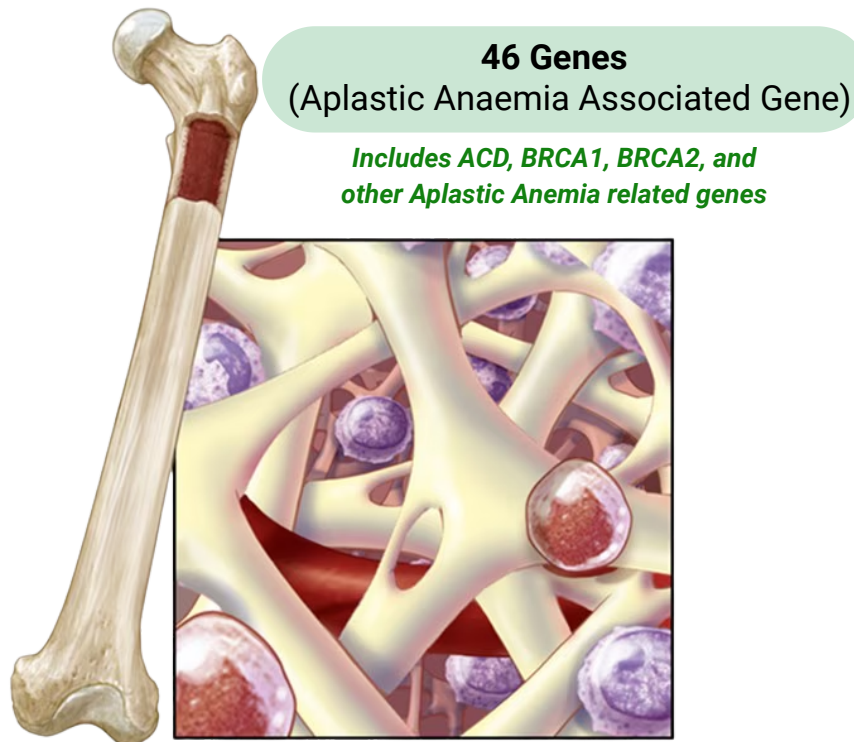


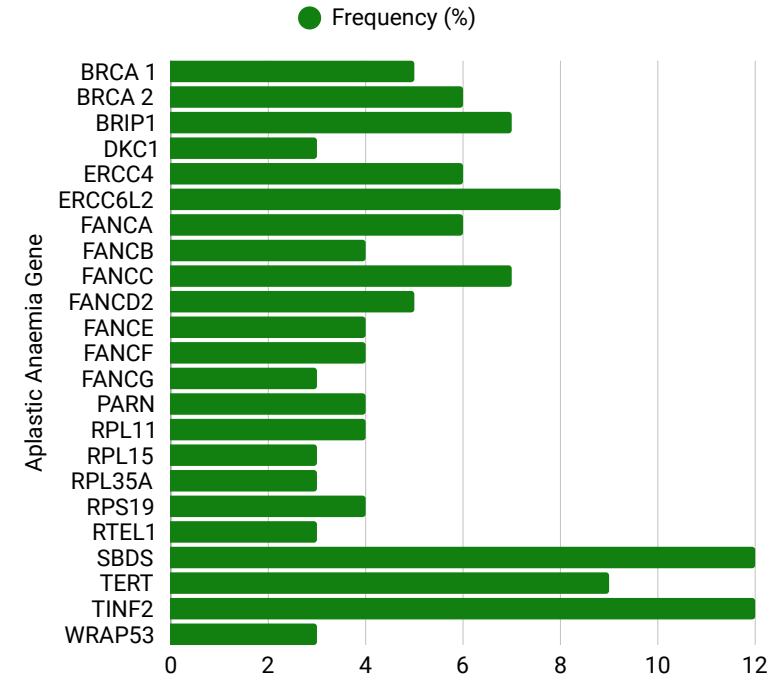
Aplastic Anaemia Gene Panel

Next Generation Sequencing based test to
detect Genomic Biomarker



Aplastic Anaemia Gene Frequency Table

Summary of Gene frequencies in Aplastic Anaemia based on dataset



Key Insights (Clinically Relevant)

Top Yield Genes (Priority Testing)

- TINF2 (~12–13%)
- SBDS (~12%)
- TERT (~9%)
- ERCC6L2 (~8%)
- BRIP1 / FANCC (~7%)

Biological Pathway Clusters

DNA Repair / Fanconi pathway (major chunk)
FANCA, FANCC, FANCD2, BRCA1/2, BRIP1
Telomere biology
TERT, TINF2, RTEL1, DKC1
Ribosomal disorders
RPL & RPS genes (Diamond-Blackfan overlap)
Other hematopoietic regulators
GATA1, SBDS

Gene List

Aplastic Anaemia associated genes

ACD, BRCA1, BRCA2, BRIP1, DKC1, ERCC4, ERCC6L2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, GATA1, MAD2L2, NBN, NHP2, NOP10, PALB2, PARN, PRF1, RAD51, RAD51C, RPL11, RPL15, RPL26, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS28, RPS29, RPS7, RTEL1, SBDS, SLX4, TERT, TINF2, TSR2, UBE2T, WRAP53

Test Details

Test Code	MGM042
Test Name	Aplastic Anaemia Gene Panel
TAT	19 days
Specimen Type	Peripheral Blood in EDTA (Purple Top) 20-25°C, Chorionic Villus Sample (CVS)/-2-8°C, Direct DNA/-20-25°C, Amniotic fluid (AF)/-20-25°C
Accreditation	CAP, NABL

MedGenome

MedGenome is a global leader in genetic testing, genomics research, and drug discovery, delivering precise genomic solutions for better health management.

As the only CAP-accredited genetic testing lab in India, MedGenome offers over 1,300 advanced tests using cutting-edge technology.

Their mission is to enable faster, smarter diagnosis of complex diseases while making high-quality, affordable genetic testing accessible to over 4 billion people worldwide.

Witty Charman CoTS

Focused on Medical Technology especially in Forensic Pathology & Forensic Science related product & services. It is venturing into Diabetes Care & Digital Pathology.

Witty Charman Medical

Started with a vision to make a difference in people's health & lives through affordable, brilliant & compassionate patient care, of all age groups from pediatric to geriatric,

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Witty Charman Medical



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