

GENETIC TESTING

Under the direction of Dr Karl Baumgart, Sonic Clinical Institute performs a range of genetic assays in the Division of Molecular Genetics including DNA testing for parentage, relationship and immigration. In conjunction with Douglass Hanly Moir Pathology, SCI offers the following range of genetic assessments:

■ Alpha-Thalassaemia	■ Familial Mediterranean Fever (FMF)
■ Asthma (Beta-2 Adeno receptor Mutation)	■ Familial Hibernian Fever (TRAPS)
■ Beta-Thalassaemia Sequencing	■ Fragile-X
■ C-1 Esterase Sequencing	■ Haemochromatosis
■ Coeliac HLA Typing DR/DQ	■ Hereditary Angioedema (Type 111 HAE)
■ Connexin-26	■ HLA B27
■ Cystic Fibrosis Delta F 508	■ HLA B1502
■ Cystic Fibrosis 32 mutation screen	■ HLA B5701
■ DAZ	■ JAK 2
■ DNA Relationship Testing	■ MTHFR
■ Factor 2 (Prothrombin Gene Mutation)	■ TPMT
■ Factor 5 Leiden	



SONIC
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REFERENCES

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PRENATAL TESTING

- Sonic Clinical Institute is NATA accredited to AS/NZS 4633: 2004 (ISO 15189: 2003)
- High quality laboratory systems
- Rapid turnaround of results
- Tailored services



This document is issued in accordance with the NATA RCPA accreditation requirements.
NATA Accreditation No. 2178



FOR OPTIMAL PRENATAL CARE

WHO WE ARE

Sonic Clinical Institute (SCI), is a specialised reference facility, consisting of laboratories that perform complex and esoteric testing. SCI is NATA accredited to AS/NZS 4633: 2004 (ISO 15189: 2003), and is situated at Macquarie Park in Sydney.

SCI is part of the Sonic Healthcare group of diagnostic practices. Sonic Healthcare is an independent, Australian owned, publicly listed company, which operates in the field of medical diagnostics.

SCI offers a service of the highest quality, utilising advanced equipment and technologies. Our team of specialist pathologists and expert scientists are readily available for consultation and advice on all aspects of our testing.

DR MICHAEL J SINOSICH

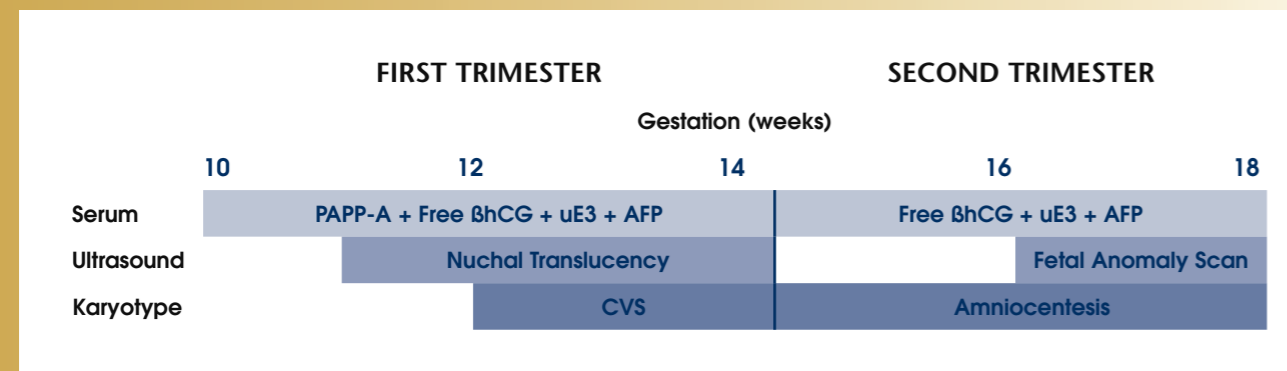
Scientific Director – Prenatal Testing

Dr Michael J Sinosich established the Division of Prenatal Testing within Sonic Clinical Institute in 2002. Michael is an internationally respected investigator in early pregnancy well-being. He has published extensively in peer reviewed journals and presented numerous papers at local, national and international forums.

The focus of Michael's research has been his work on Pregnancy-Associated Plasma Protein-A (PAPP-A). Michael launched his investigations on PAPP-A in 1980 and his early clinical studies established PAPP-A as a biochemical marker with immense clinical potential¹⁻³. These studies have subsequently been independently substantiated and his vision of early pregnancy screening is now an established reality.

TIME LINE AND TEST OPTIONS

At Sonic Clinical Institute, we have established a seamless First and Second Trimester Screening program. This has obvious advantages in those situations where patients are uncertain of dates.



Tests and time frames for the assessment of pregnancy well-being.

PRENATAL SCREENING USING SERUM MARKERS

Risk assessment is based upon the difference observed in serum marker levels between affected and unaffected pregnancies at the same gestational age. Although the difference between single biochemical marker levels may be small, the use of multiple markers greatly enhances the ability to identify pregnancies that may be at risk.

Serum marker levels change during the course of pregnancy⁴⁻⁶. Therefore, it is important that gestational age is precise to ensure that accurate Multiple of Median (MoM) values are obtained for your patient.

BIOCHEMICAL MARKERS USED FOR SCREENING

For First Trimester Screening, the biochemical markers that are measured consist of Pregnancy-Associated Plasma Protein-A (PAPP-A), Free Beta subunit of human Chorionic Gonadotrophin (FB-hCG), unconjugated Estriol (uE3) and Alpha-fetoprotein (AFP)⁷.

For Second Trimester Screening, the "Triple Test" is performed using FB-hCG, uE3 and AFP.

PRENATAL TESTING OPTIONS

When considering prenatal testing for your patient, the available options include:

- First Trimester Screening – Biochemistry with Nuchal Translucency (NT)
- Second Trimester Screening – Biochemistry
- Integrated Test – First and Second Trimester Screening performed sequentially

SCI offers established programs for both First and Second Trimester Screening.

OPTIMAL TIME FOR TESTING

Serum collected during either the first or second trimester can be used to determine the likelihood of Down's syndrome and other chromosomal anomalies such as Edward's syndrome (Trisomy 18).

Testing during the first trimester, however, is optimal for detecting autosomal aneuploidies or compromised early pregnancy.

Second trimester testing between 15 to 17 completed weeks (17 weeks 6 days) gestation is optimal for the assessment of structural anomalies such as neural tube defect (NTD).

For optimal prenatal care, First and Second Trimester Screening may be considered to be complementary.

SERUM TESTING AND NUCHAL TRANSLUCENCY

Much has been made of the "one-stop shop" model for First Trimester Screening. Whilst this model may suit some practices, it does not necessarily confer optimal diagnostic performance.

For detection of Trisomy 21, maximal diagnostic efficiency for PAPP-A is achieved at 10 to 11 completed weeks (11 weeks 6 days) gestation⁸. Therefore, it is quite acceptable to measure biochemical markers in advance of the nuchal scan that is normally performed at 11 to 13 completed weeks (13 weeks 6 days) gestation.

With our rapid turn-around time (daily Sunday to Friday), first trimester MoM values of serum markers are available for incorporation into risk assessment software well in advance of the patient's appointment for ultrasound.

SPECIMEN REQUIREMENTS

- Collect anticoagulant free blood (10 mL)
- For short-term storage and transport (less than 6 hours) – separate and refrigerate serum (4-8°C)
- For long-term storage and transport (more than 6 hours) – separate and freeze serum

COLLECTION FACILITIES

- For location of collection centres nationwide:
 - Please phone Toll Free 1800 004 052 or
 - View our website: www.sonicclinicalinstitute.com.au
- Appointments are not necessary

REPORT OPTIONS

Several reporting options are available, for example:

- Electronic download
 - Fax or telephone
 - Hard copies delivered via our courier network
- These may be customised to meet your individual needs. To personalise the issuing of reports, please contact our Administrative Officer on Toll-Free 1800 004 052.

REPORT UPDATES

Marker MoM recalculation and report updates are available in real-time (Toll Free 1800 004 052) and/or by routine reporting (fax, hard copy, electronic download).