PREGNARISK

Genetic Screening Test

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PANEL 1

Couples who come for genetic consultation before marriage or pregnancy can use this panel.

Panel Content

- Chromosome Analysis
- Alpha and Beta Thalassemia and Sickle Cell Anemia
- Fragile X
- Spinal Muscular Atrophy (SMA)
- Duchenne Muscular Dystrophy (DMD)
- Phenylketonuria
- Biotinidase Deficiency
- 21 Hydroxylase Deficiency
- Cystic fibrosis

Required material

- 4 cc of EDTA Blood Sample and 2 cc of Heparinized blood Sample from spouses

PANEL 2

It is a comprehensive panel that can be recommended during or before pregnancy for couples with or without consanguineous marriage. It includes a significant part of the diseases with a frequency of 1/200 and above that are "recommended" based on ACMG criteria. More than 4000 diseases that cause risks to maternal and infant health are screened during pregnancies.

Panel Content

- Chromosome Analysis
- Clinical Exome Sequencing (CES)
- Fragile X
- Spinal Muscular Atrophy (SMA)
- Duchenne Muscular Dystrophy
- 21 Hydroxylase Deficiency

Required material

- 4 cc of EDTA Blood Sample and 2 cc of Heparinized blood Sample from spouses

PANEL 3

This panel should be the first panel to be recommended for consanguineous couples. Since this panel reduces the risk very much, it can also be recommended for couples who do not have consanguineous marriages. A huge amount of data is gained in panels 2 and panel 3. This information can sometimes show the signs of a disease that exists in the person and has a treatment. Based on this, if the family requests, an evaluation can be made from this point of view. About 8000 diseases that cause risks to maternal and infant health are screened during pregnancies.

Panel Content

- Chromosome Analysis
- Whole Exome Sequencing (WES)
- Fragile X
- Spinal Muscular Atrophy (SMA)
- Duchenne Muscular Dystrophy (DMD)
- 21 Hydroxylase Deficiency

Required material

- 4 cc of EDTA Blood Sample and 2 cc of Heparinized blood Sample from spouses

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