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Microreader™ Human SMN1 gene exon deletion Detection Kit

Spinal muscular atrophy (SMA) is the most common fatal autosomal recessive genetic disease in infants and young children, with a carrier rate of about 1/50 in the population. The clinical manifestations are progressive, symmetrical, proximal limb muscle weakness and muscle atrophy, and are divided into types 0-IV according to the time of onset and clinical manifestations. About 95% of SMA patients are caused by homozygous deletion of exon 7 of SMN1, and the copy number of SMN2 affects the prognosis of the disease.

The kit uses fluorescent PCR-capillary electrophoresis to detect the copy numbers of SMN1 and SMN2 and the SNP loci related to "2+0" carriers, which can accurately distinguish healthy people, carriers and patients, and is used for SMA carriers Screening and auxiliary diagnosis of patients.

Features

Accurate results	The kit is stable and can accurately distinguish healthy people, carriers, and patients
Comprehensive detection	Can detect SMN1, SMN2 copy number and "2+0" carrier-related point mutations
Simple and fast operation	Based on fluorescent PCR-capillary electrophoresis, the whole detection process takes about 4 hours

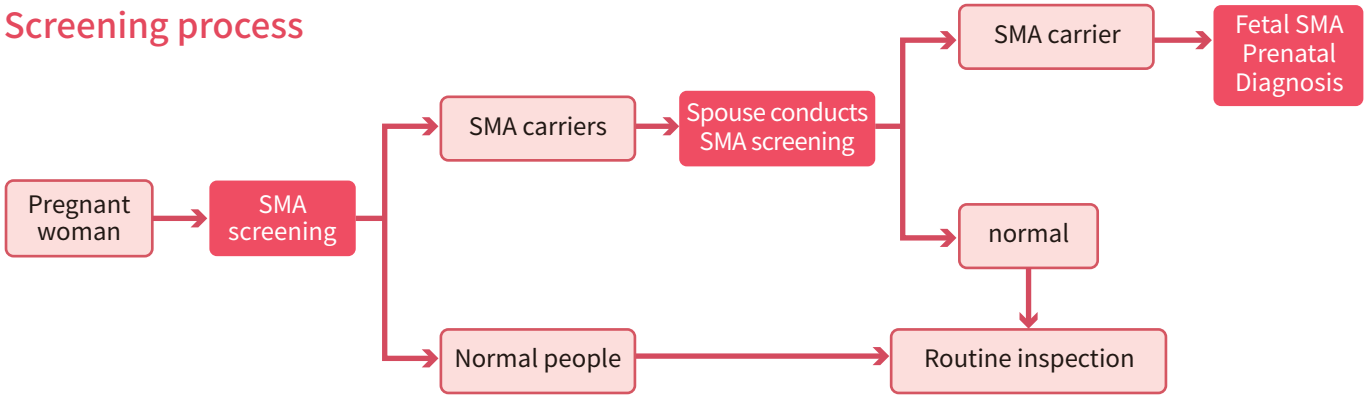
For people

All couples who are trying to conceive or are pregnant are eligible for SMA carrier screening.

SMA suspected individuals with associated symptoms.

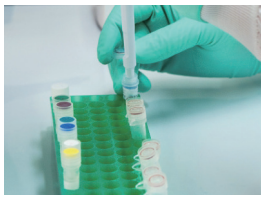
- In 2008, the American Medical Genetics Association (ACMG) recommended pan-ethnic screening for SMA, that is, regardless of region or race, SMN1 carrier genetic screening should be implemented for all gestational age groups without distinction, and prenatal screening for high-risk fetuses should be carried out. diagnosis, thereby reducing the number of births of children with SMA.
- In 2017, the American College of Obstetricians and Gynecologists (ACOG) recommended SMA screening for all women planning or already pregnant.

Screening process



Detection process

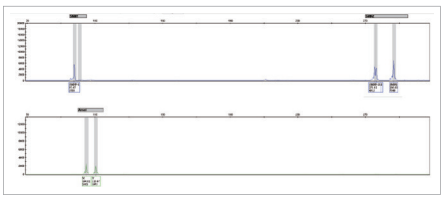
1 DNA extraction



2 Fluorescent PCR amplification

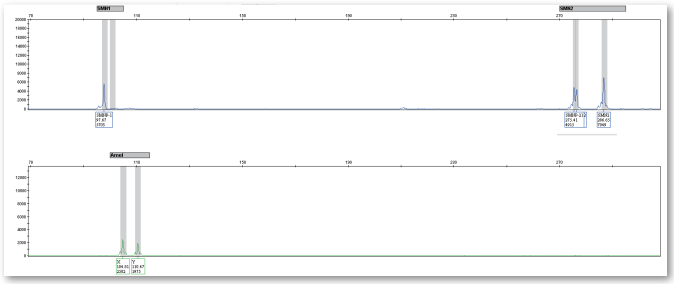
3 Detection on Capillary electrophoresis platform

4 Results interpretation

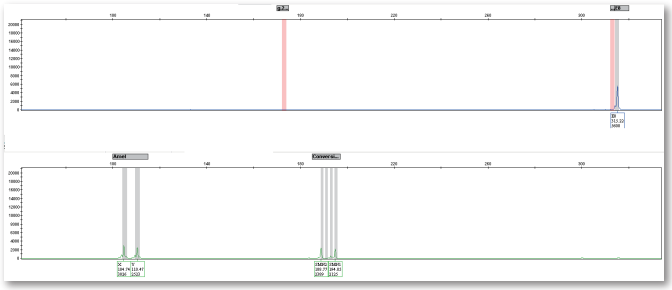


Results

CNV system



SNP system



Applicable instrument

PCR instrument: Life Technologies Holdings Pte Ltd: 9700, Veriti DX;
Genetic analyzer: Life Technologies Holdings Pte Ltd: 3730xL Dx, 3500 Dx; Seqstudio.

Sampling requirements

- EDTA anticoagulant, genomic DNA

Kit specification

- 24 reactions / kit, 48 reactions / kit

※ This product is only for scientific research use, and this information is only for reference by relevant medical professionals. Please refer to the instruction manual for details of contraindications or precautions.

WeChat public account

MRC-SMN1-23-1