

Direct detection PGT-M for Spinal Muscular Atrophy (SMA) — Now Available at Orchid

What is SMA?

Spinal Muscular Atrophy (SMA) is a serious autosomal recessive neurodegenerative condition which causes severe muscle weakness and atrophy and can result in death before the age of 2 in severe cases. It is most commonly caused by deletions within the *SMN1* gene.

Who's at Risk?

- About 1 in 40 people are carriers of SMA.
- Some people are called “silent carriers” who carry two copies on one chromosome and none on the other (called a 2+0 carrier)
 - Silent carriers may be at high risk for an affected offspring if their partner is a confirmed or silent carrier
 - Standard PGT-M platforms can't accommodate this complex situation, leaving patients with no options for risk reduction in embryos

The Solution: Orchid's SMA Screening with PGT-M

Orchid now offers targeted embryo screening (PGT-M) for couples at risk for SMA; even in complicated cases, without the need for custom tools or extended family studies.

How?

Orchid provides direct testing for *SMN1** gene copies in embryos to report those with zero copies (affected) vs those with one or more (unaffected)

Why This Matters

Until now, families with silent carrier risk often had no way to screen embryos for SMA. Enter Orchid

Additional Benefits with Orchid's PGT-WGS:



Patients needing PGT-M for SMA have the added benefit of screening for hundreds of other serious genetic conditions



Simplified process — no need to create a custom probe or gather family samples



Faster turnaround time than traditional methods

Available Now

Orchid's PGT-WGS with SMA add-on is available for:

- Couples who are confirmed carriers of SMA
- Couples with suspected increased risk (including silent carrier concerns)



Email genetics@orchidhealth.com or schedule a call using the QR code here to learn how you can offer Orchid's screening to your patients!



*Please note that sequence variants will not be reported

**~2% of SMA cases are the result of a de novo pathogenic variant not detected by Orchid's PGT-M for SMA