

DMD Deletion/Duplication Detection

NGS data for Otogenetics Carrier Screen (ACOG 13/14 genes) was used for CNV analysis.
Samples analyzed: 2786

Male samples analyzed: 214; 100% identified as having one copy of DMD

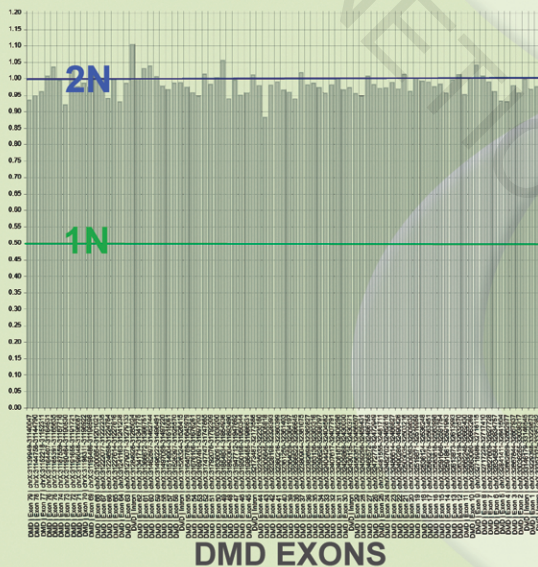
Coriell known DMD del/dup samples: 13; 100% identified correctly*

*The described del/dup of 3 Coriell samples was not accurate. Otogenetics CNV detected DMD del/dup was confirmed by array analysis.

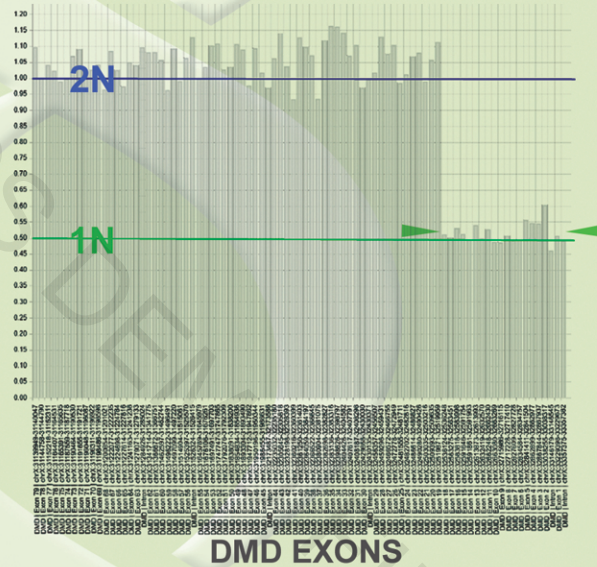
DMD del/dup positive samples identified: 7; all confirmed by array analysis.

Copy Numbers Based on Normalized Coverage

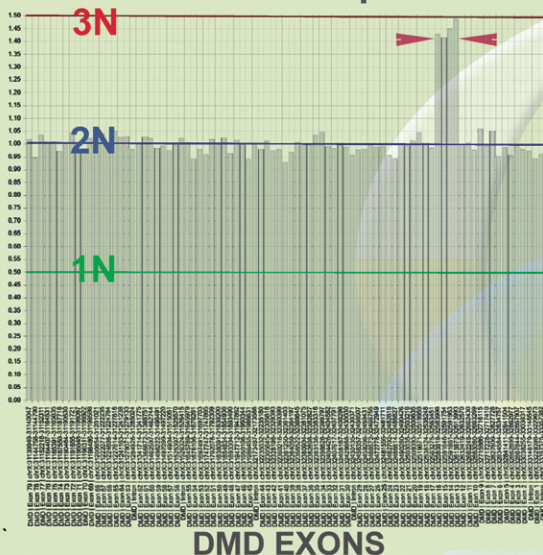
Normal Control



Coriell Known DMD Deletion



Detected DMD Duplication



Detected DMD Deletion

