SMA is the most common neurodegenerative disease in childhood with an incidence of 1 in 6,000 to 1 in 10,000. SMA is caused by deleterious changes in the SMN1 gene, with a deletion of exon 7 being the most common pathogenic event. Homozygous deletion of exon 7 can be found in approximately 95% of SMA cases, whereas the other 5% are compound heterozygous of this deletion.

- **Fragment analysis using the AmpliDex® PCR/CE SMN1/2 Plus Kit.** The assay is based on PCR and capillary electrophoresis. In addition to SMN1/2 copy number determination, the assay also detect the presence/absence of gene duplication masks and modifier c.*3+80T>G, c.*211_*212del, and c.859G>C.

- **Identification of SMA positive (SMN1=0) and SMN1 carrier statues using WGS data.** A bioinformatic workflow was developed and validated for the SMN1 copy number determination through uniquely mapped reads on exon 7 of SMN1 gene using the WGS data. Median read depth of SMN1/SMN2 exon 7 for WGS samples from previous runs are computed and used for normalization.

### CONCLUSION

- Currently, this laboratory performs qPCR assay for population-based newborn screening for SMA, bioinformatic analysis of WGS data for identification of SMA patients (SMN1=0) and SMN1 carrier statues, fragment analysis and MLPA for confirmation and diagnostic testing for determination of SMN1/2 copy numbers.
- The combination of above assays reduce the time to diagnosis of SMA.