GENETIC TEST:
Congenital Central Hypoventilation Syndrome / Ondine syndrome

FULL NAME:	Congenital Central Hypoventilation Syndrome / Ondine syndrome
DESCRIPTION:	1) To detect non-polyalanine repeat expansion mutations (NPARMs), and polyalanine repeat expansion mutations (PARMs), Sanger sequencing of the amplicons is performed. Large copy number variations (deletions, insertions, duplications) cannot be excluded with this analysis. 2) PHOX2B has two polyalanine repeat regions in exon 3. Therefore the amplicons are also analysed by capillary electrophoresis on a Fragment Analyzer device. In most CCHS individuals with neonatal onset, polyalanine repeat expansion mutations (PARMs) of 25 or more GCN repeats can be found (the N in GCN represents any nucleotide: A/C/G/T). Also the 35-bp and 38-bp NPARM recurrent out-of-frame deletions within the GCN repeat region can be detected. 3) As PHOX2B partial and whole-gene deletions have also been reported, MLPA analysis (multiplex ligation dependent probe amplification) is performed for detection of copy number variations (CNVs) in the PHOX2B gene (MRC Holland, P318-Hirschsprung-2, version A2).
TEST TYPE:	Clinical

TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Post-natal Diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA
METHOD CATEGORY:	Sequence analysis: entire coding region Deletion/duplication analysis
METHOD TECHNIQUE:	Next Generation Sequencing (NGS) MLPA based techniques
RIZIV CODE:	565471-565482
TURNAROUND TIME (MAXIMUM):	3-4 months
CREATED:	29 Jul 2019 - 13:49
CHANGED:	04 Dec 2023 - 16:58

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