CENTOGENE
THE RARE DISEASE COMPANY

**CENTOGENE AG** Am Strande 7 • 18055 Rostock • Germany

Name
Hospital
Address

**Order no.:** xxxxxx
**Order received:** xxxxxx
**Sample type:** xxxxxx
**Sample collection date:** xxxxxx
**Report date:** xxxxxx

Patient no.: **xxxxxx**, First Name: **xxxxxx**, Last Name: **xxxxxx**
DOB: **xxxxxx**, Sex: **xxxxxx**, Your ref.: **xxxxxx**

**Test(s) requested: carrier testing in the NSD1 (OMIM®: 606681) gene**

## RECLASSIFICATION REPORT

Based on new evidences becoming available for the NSD1 (NM_022455.4) variant c.6557A>G p.(His2186Arg) we have reclassified this variant from uncertain clinical significance (class 3) to likely pathogenic (class 2).

### REASON FOR RECLASSIFICATION

We previously detected this variant in a heterozygous state in this patient. ClinVar lists this variant as likely pathogenic (clinical testing, Variation ID: 159430). As parental carrier testing has shown that this variant is *de novo* in this patient, and the variant has strong *in silico* predictions and is absent in external population databases, we have reclassified this variant.

**Therefore, in your patient, this result is likely consistent with a genetic diagnosis of Sotos syndrome type 1.**

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## CENTOGENE VARIANT CLASSIFICATION (BASED ON ACMG RECOMMENDATIONS)

**Class 1** – Pathogenic
**Class 2** – Likely pathogenic
**Class 3** – Variant of uncertain significance (VUS)
**Class 4** – Likely benign
**Class 5** – Benign

Additionally, other types of clinical relevant variants can be identified (e.g. risk factors, modifiers).

## ADDITIONAL INFORMATION

This test was developed and its performance validated by Centogene AG. The US Food and Drug Administration (FDA) has determined that clearance or approval of this method is not necessary and thus neither have been obtained. This test has been developed for clinical purposes. All test results are reviewed, interpreted and reported by our scientific and medical experts.

## DISCLAIMER

Any preparation and processing of a sample from patient material provided to CENTOGENE by a physician, clinical institute or a laboratory (by a "Partner") and the requested genetic and/or biochemical testing itself is based on the highest and most current scientific and analytical standards. However, in very few cases genetic or biochemical tests may not show the correct result, e.g. because of the quality of the material provided by a Partner to CENTOGENE or in cases where any test provided by CENTOGENE fails for unforeseeable or unknown reasons that cannot be influenced by CENTOGENE in advance. In such cases, CENTOGENE shall not be responsible and/or liable for the incomplete, potentially misleading or even wrong result of any testing if such issue could not be recognized by CENTOGENE in advance.

## COPYRIGHT NOTICE:



Chief Medical Director

Clinical Scientist

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