NEURAZON
WELLNESS STARTS WITH YOU

From:
Neurazon INC.
Precision Health Analysis Department - Children with Autism and Developmental Disorders
Quebec, Canada

Case no: 5102415365
First Name: Rahaf
Last Name: Khaled
DOB: 20.05.2022
Sex: Female

Test requested: Precision Health Analysis®

Results:
True Autism Spectrum Disorder Variants: NEGATIVE
Acquired Autism Spectrum Disorder Variants: POSITIVE
Attention-Deficit/Hyperactivity Disorder Variants: NEGATIVE
Information Processing and Brain Development Variants: NEGATIVE
White Matter Delays Variants: POSITIVE
Methylation Variants: NEGATIVE
Metabolic and Mitochondrial Function Variants: NEGATIVE
Neurotransmitters, Synaptic Health, and Behavior Variants: POSITIVE
Immune System Variants: NEGATIVE
Potential Digestive System Variants: NEGATIVE
Epigenetic Regulation Variants: NEGATIVE
Potential Uncontrolled Electrical Activity: NEGATIVE

Recommended Interpretation:

- Please note any clinically relevant variants detected that are associated with the described phenotype.
- Review all variants listed in the attached report, if applicable.
- We recommend reevaluating the sequence dataset every 12 months or when there are changes in the phenotype.
- Reassessment by a multidisciplinary team is advised every 3 months to monitor any observed phenotypic changes using artificial intelligence.
- A tailored intervention plan should be developed and supervised by a multidisciplinary team.

ADDITIONAL INFORMATION

This assessment has been developed and validated through the AI-powered bioinformatics platform of NEURAZON, which integrates advanced data analysis with input from partner laboratories worldwide. All results are carefully reviewed, interpreted, and compiled by NEURAZON's team of scientific experts. For full details, please refer to the attached report.

> Contact Details

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Recommended Homeopathy Plan

Step: 1st step
Name: Rahaf Khaled
DOB: 20.05.2022

Please refer to external report

ADDITIONAL INFORMATION

Recommendations: The provided guidance includes safe, non-drug, natural treatments intended to support the child's development. These recommendations are not a replacement for professional rehabilitation and training sessions and should always be followed under the supervision of a qualified specialist. Progress and improvement depend on the individual child, the underlying causes, and their response to the interventions, with recommendations adjusted based on regular evaluations every three months. If any signs of an allergic reaction occur, discontinue the treatment immediately and consult with a healthcare provider to modify the treatment plan accordingly.

Reevaluation: A reevaluation is necessary after the specified duration to reassess development using the AI model or as recommended by the professional team. This test doesn't replace a full clinical assessment by a physician.

Next Steps: Begin the second phase of the protocol after the time-frame indicated in the treatment plan.

This assessment has been developed and validated through the AI-powered bio-informatics platform of NEURAZON, which integrates advanced data analysis with input from partner laboratories worldwide. All results are carefully reviewed, interpreted, and compiled by NEURAZON's team of scientific experts. For full details, please refer to the attached report. Neurazon is a bio-informatics platform that analyzes data, not a laboratory; however, it partners with leading international laboratories to ensure the highest level of accuracy.

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Case no: 5102415365
First Name: RAHAF
Last Name: KHALED
DOB: 20.05.2022
Sex: Female

Order no: 5102415365
Report Type: Final report

Requested Test: WES (Whole Exome Sequencing)
Clinical Information/Indication: The analysis was performed according to the demand of referal center to identify variants in relevant genes associated with neurological disorders.
Result:

Main Findings: **A likely pathogenic variant; c.1283G>C / p.Arg428Thr in the KATNAL2 gene was identified.**					
Gene (Transcript)	**Location**	**Nucleotide (Protein) dbSNP**	**Zygosity**	**Variant Classification**	**Disease (OMIM#, Inheritance)**
KATNAL2 NM_001387690.1	Exon 16	c.1283G>C p.Arg428Thr	Heterozygous	Likely Pathogenic	Autism Related Disorder[2] Unknown Inheritance

Details About Gene and Variants:
KATNAL2: Predicted to enable ATP hydrolysis activity. Predicted to be involved in cytoplasmic microtubule organization. Located in cytoplasm; microtubule; and spindle pole. [provided by Alliance of Genome Resources, Jul 2025]

The KATNAL2 variant c.1283G>C / p.Arg428Thr was detected in heterozygous state. It is classified as Likely Pathogenic according to the recommendations of ACMG. This variant is associated with Autism Related Disorder[2].

[2] Sanders, Stephan J et al. “De novo mutations revealed by whole-exome sequencing are strongly associated with autism.” Nature vol. 485,7397 237-41. 4 Apr. 2012, doi:10.1038/nature10945

Recommendations:
- It is recommended to evaluate this patient’s result together with clinical and laboratory findings.
- Genetic screening and clinical evaluation are recommended for family members.
- Genetic counseling is recommended.

Additional Findings:
The variants reported as additional findings refer to pathogenic changes that are not related to the referred phenotype but can cause additional phenotype thus require genetic counseling and further clinical evaluation.

ACMG Findings (ACMG 81 Genes):
According to the ACMG guidelines (Genetics in Medicine, 2023; PMID: PMID: 37347242), pathogenic and likely pathogenic incidental variants in proposed diseases and genes are reported. Variants in the BRCA1/2, MLH1, MSH2, MSH6, PMS2, TMEM127, MAX and MUTYH genes associated with adult onset diseases in children (under 15 years of age) are not reported.

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Order no: 5102415365
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Case no: 5102415365
First Name: RAHAF
Last Name: KHALED
DOB: 20.05.2022
Sex: Female

Carriership Findings:
Pathogenic and likely pathogenic variants in genes selected according to OMIM phenotypes associated with severe or early-onset diseases have been reported. The variants reported as carriership findings refer to pathogenic changes that are not related to the reported phenotype and require family screening in conjunction with genetic counseling due to potential carrier risk.

Gene (Transcript)	Location	Nucleotide (Protein) dbSNP	Zygosity	Variant Classification	Disease (OMIM#, Inheritance)
BCS1L NM_001079866.2	Exon 4	c.550C>T p.Arg184Cys rs121908578	Heterozygous	Pathogenic	BCS1L Related Disease OMIM: 603647 Autosomal Recessive

CNV Analysis:

CNV Detected Genes and Region	Result	Disease (OMIM#, Inheritance)
MEFV (Exons 1-10) chr16:3.293.131-3.306.598 (NGS)	Pathogenic, Heterozygous Deletion chr16:3.293.131-3.306.598 16p13.3 (13.5kb) sseq[GRCh37] DEL(chr16)(16p13.3) (3293131_3306598)	Familial Mediterranean Fever OMIM: 249100 Autosomal Recessive

Method:
Whole Exome Sequencing is performed by DiagnoSeq using Twist Biosciences technology. First, approximately 36.5 Mb of Consensus Coding Sequences (CCS) (targeting> 98% of RefSeq and Gencode v28 regions derived from the human genome) are replicated from fragmented genomic DNA with the Twist Human Core Exome Plus kit. The generated library is sequenced on the Illumina Novaseq NGS platform to achieve a minimum reading depth of 20x for>98% of the targeted bases. As a result of sequencing, raw data is obtained in FASTQ and VCF formats. Whole Exome Sequencing analysis is performed on FASTQ data using the Franklin by Genoox analysis program. In addition to all disease-causing variants reported in the HGMD®, ClinVar, and CentoMD® databases as well as all variants with a Minor Allele Frequency (MAF) of less than 1% in the gnomAD database are considered. The research for related variables has focused on coding exons and surrounding +/- 20 intronic bases. All potential inheritance patterns are covered. In addition, the family history and clinical information provided are used to evaluate pathogenicity and variables defined by their cause of disease. Sequencing variants are classified into five classes according to the ACMG/AMP variant classification guidelines and ClinGen recommendations: pathogenic, likely pathogenic, Variant of unknown significance (VUS), likely benign, and benign[1]. All variables related to the patient's phenotype are reported, except for benign or possible benign variants. Low-quality single nucleotide variants and all related deletion/insertion variants are validated by Sanger sequencing.

Analysis Statistics:

Average Depth	148
Variant Quality	99%

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