## Nathalie Boddaert

## List of Publications by Year in descending order

Source: https:|/exaly.com/author-pdf/7655984/publications.pdf
Version: 2024-02-01


| 1 | Targeted therapy in patients with PIK3CA-related overgrowth syndrome. Nature, 2018, 558, 540-546. | 13.7 | 374 |
| :---: | :---: | :---: | :---: |
| 2 | Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. Nature Genetics, 2017, 49, 1529-1538. | 9.4 | 164 |
| 3 | Type I interferon-mediated autoinflammation due to DNase II deficiency. Nature Communications, 2017, 8, 2176. | 5.8 | 164 |
| 4 | Clinical and imaging diagnosis for heredodegenerative diseases. Handbook of Clinical Neurology \| Edited By P J Vinken and G W Bruyn, 2013, 111, 63-78. | 1.0 | 124 |
| 5 | Loss-of-Function Mutations in WDR73 Are Responsible for Microcephaly and Steroid-Resistant Nephrotic Syndrome: Galloway-Mowat Syndrome. American Journal of Human Genetics, 2014, 95, 637-648. | 2.6 | 108 |
| 6 | Functional neuroimaging and childhood autism. Pediatric Radiology, 2002, 32, 1-7. | 1.1 | 107 |
| 7 | Mutations in QARS, Encoding Clutaminyl-tRNA Synthetase, Cause Progressive Microcephaly, Cerebral-Cerebellar Atrophy, and Intractable Seizures. American Journal of Human Genetics, 2014, 94, 547-558. | 2.6 | 106 |

8 Reverse-Transcriptase Inhibitors in the Aicardiâ€"GoutiÃ"res Syndrome. New England Journal of
Histone H3 wild-type DIPG/DMG overexpressing EZHIP extend the spectrum diffuse midline gliomas with
9 PRC2 inhibition beyond H3-K27M mutation. Acta Neuropathologica, 2020, 139,1109-1113.
Clinical, laboratory and molecular findings and long-term follow-up data in 96 French patients with

10 | PMM2-CDG (phosphomannomutase 2-congenital disorder of glycosylation) and review of the |
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literature. Journal of Medical Genetics, 2017, 54, 843-851.
$\left.\begin{array}{ll}\text { Long-Term Outcome of } 106 \text { Consecutive Pediatric Ruptured Brain Arteriovenous Malformations After } \\ \text { Combined Treatment. Stroke, 2014, 45, 1664-1671. }\end{array}\right]$
12 Biallelic Mutations in MRPS34 Lead to Instability of the Small Mitoribosomal Subunit and Leigh ..... 2.6 ..... 83 Syndrome. American Journal of Human Genetics, 2017, 101, 239-254.
3.6 ..... 82Arterial Spin Labeling to Predict Brain Tumor Grading in Children: Correlations between
Histopathologic Vascular Density and Perfusion MR Imaging. Radiology, 2016, 281, 553-566.Mutation in PNPT1, which Encodes a Polyribonucleotide Nucleotidyltransferase, Impairs RNA Import14 into Mitochondria and Causes Respiratory-Chain Deficiency. American Journal of Human Genetics,2.6
2012, 91, 912-918.
15 Defects in t6A tRNA modification due to GON7 and YRDC mutations lead to Galloway-Mowat syndrome.
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20

A nonsense variant in HERC1 is associated with intellectual disability, megalencephaly, thick corpus callosum and cerebellar atrophy. European Journal of Human Genetics, 2016, 24, 455-458.
1.4

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From splitting GLUT1 deficiency syndromes to overlapping phenotypes. European Journal of Medical Genetics, 2015, 58, 443-454.
21

Myocardial inflammation detected by cardiac MRI in Arrhythmogenic right ventricular
0.8
cardiomyopathy: A paediatric case series. International Journal of Cardiology, 2018, 271, 81-86.
52

Early epileptic encephalopathies associated with STXBP1 mutations: Could we better delineate the
0.7

50
phenotype?. European Journal of Medical Genetics, 2014, 57, 15-20.
High-grade gliomas in adolescents and young adults highlight histomolecular differences from their
adult and pediatric counterparts. Neuro-Oncology, 2020, 22, 1190-1202.
$0.6 \quad 50$

Inherited deficiency of stress granule ZNFX1 in patients with monocytosis and mycobacterial disease.
Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .
3.3
25 Arterial Spin Labeling MRI: A step forward in non-invasive delineation of focal cortical dysplasia in children. Epilepsy Research, 2014, 108, 1932-1939.
Mutations in DOCK7 in Individuals with Epileptic Encephalopathy and Cortical Blindness. American Journal of Human Genetics, 2014, 94, 891-897.$0.8 \quad 46$
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28
Cerebral Blood Flow Improvement after Indirect Revascularization for Pediatric Moyamoya Disease: A

Statistical Analysis of Arterial Spin-Labeling MRI. American Journal of Neuroradiology, 2016, 37, 706-712.

Brain imaging in mitochondrial respiratory chain deficiency: combination of brain MRI features as a
useful tool for genotype/phenotype correlations. Journal of Medical Genetics, 2014, 51, 429-435.
1.5

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29 Multiple bur hole surgery for the treatment of moyamoya disease in children. Journal of
Neurosurgery: Pediatrics, 2006, 105, 437-443.

0.8

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Moyamoya syndrome in children with neurofibromatosis type 1: Italianấ"French experience. American
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Myocardial involvement in children with post-COVID multisystem inflammatory syndrome: a
32 cardiovascular magnetic resonance based multicenter international studyâ $€$ "the CARDOVID registry.
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Journal of Cardiovascular Magnetic Resonance, 2021, 23, 140.
33 <i>|FT81<|i>, encoding an IFT-B core protein, as a very rare cause of a ciliopathy phenotype. Journal of Medical Genetics, 2015, 52, 657-665.
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Further refinement of COL4A1 and COL4A2 related cortical malformations. European Journal of
Medical Genetics, 2018, 61, 765-772.

WDR81 mutations cause extreme microcephaly and impair mitotic progression in human fibroblasts and Drosophila neural stem cells. Brain, 2017, 140, 2597-2609.
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Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nature Communications, 2021, 12, 2558.

A partial form of inherited human USP18 deficiency underlies infection and inflammation. Journal of Experimental Medicine, 2022, 219, .
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Massive and exclusive pontocerebellar damage in mitochondrial disease and <i>NUBPL</i> mutations.
Neurology, 2012, 79, 391-391.

Treatment of two infants with PIK3CA-related overgrowth spectrum by alpelisib. Journal of
Experimental Medicine, 2022, 219, .
$43 \quad \begin{aligned} & \text { Magnetic resonance imaging arterialâ€spinâ€labelling perfusion alterations in childhood migraine with } \\ & \text { atypical aura: a caseấ } \text { écontrol study. Developmental Medicine and Child Neurology, 2016, 58, 965-969. }^{4} \text {. }\end{aligned}$
$43 \begin{aligned} & \text { Magnetic resonance imaging arterialâ€spinâ€labelling perfusion alterations in childhood migraine with } \\ & \text { atypical aura: a caseâ€ "control study. Developmental Medicine and Child Neurology, 2016, 58, 965-969. }\end{aligned}$
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Historadiological correlations in high-grade glioma with the histone 3.3 G34R mutation. Journal of
Neuroradiology, 2018, 45, 316-322.

Challenges in managing epilepsy associated with focal cortical dysplasia in children. Epilepsy
Research, 2018, 145, 1-17.

Incidental Brain MRI Findings in Children: A Systematic Review and Meta-Analysis. American Journal of
Neuroradiology, 2019, 40, 1818-1823.

47 Scurvy: A New Old Cause of Skeletal Pain in Young Children. Frontiers in Pediatrics, 2020, 8, 8.
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radiological, histopathological and molecular features as their pontine counterparts. Acta
Neuropathologica Communications, 2020, 8, 104.
An integrative radiological, histopathological and molecular analysis of pediatric pontine
histone-wildtype glioma with MYCN amplification (HGG-MYCN). Acta Neuropathologica
histone-wilatype glioma with 1 V
Communications, 2019, 7, 87 .
Loss of Function of RIMS2 Causes a Syndromic Congenital Cone-Rod Synaptic Disease with
50 Neurodevelopmental and Pancreatic Involvement. American Journal of Human Genetics, 2020, 106,
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859-871.
51 Neuroimaging evidence of brain abnormalities in mastocytosis. Translational Psychiatry, 2017, 7,
el197-el197.

High predictive value of brain MRI imaging in primary mitochondrial respiratory chain deficiency.
Journal of Medical Genetics, 2018, 55, 378-383.

Fastâ€track virtual reality for cardiac imaging in congenital heart disease. Journal of Cardiac Surgery,
2021, 36, 2598-2602.

Arterial Spin-Labeling to Discriminate Pediatric Cervicofacial Soft-Tissue Vascular Anomalies.
American Journal of Neuroradiology, 2017, 38, 633-638.

Mutations in the <i>MRPS28</i> gene encoding the small mitoribosomal subunit protein bS1m in a
56 patient with intrauterine growth retardation, craniofacial dysmorphism and multisystemic
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involvement. Human Molecular Genetics, 2019, 28, 1445-1462.
Imaging features of medulloblastoma: Conventional imaging, diffusion-weighted imaging,
perfusion-weighted imaging, and spectroscopy: From general features to subtypes and characteristics.
Neurochirurgie, 2021, 67, 6-13.
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Radiation Oncology Biology Physics, 2017, 99, 476-485.
Elevated thrombin generation in patients with congenital disorder of glycosylation and combined
coagulation factor deficiencies. Journal of Thrombosis and Haemostasis, 2019, 17, 1798-1807.
$1.9 \quad 18$

Recurrent KIF5C mutation leading to frontal pachygyria without microcephaly. Neurogenetics, 2016,
17, 79-82.
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61 Neuropsychological improvement after posterior fossa arachnoid cyst drainage. Child's Nervous
System, 2017, 33, 135-141.
$0.6 \quad 17$

Imaging Features with Histopathologic Correlation of CNS High-Grade Neuroepithelial Tumors with a <i>BCOR</i> Internal Tandem Duplication. American Journal of Neuroradiology, 2022, 43, 151-156.
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63 Risk Factors for Early Brain AVM Rupture: Cohort Study of Pediatric and Adult Patients. American
Journal of Neuroradiology, 2020, 41, 2358-2363.

Myocardial inflammation on cardiovascular magnetic resonance predicts left ventricular function 64 recovery in children with recent dilated cardiomyopathy. European Heart Journal Cardiovascular
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Imaging, 2015, 16, 756-762.

$65 \quad$| Neuroinflammatory Disease following Severe Acute Respiratory Syndrome Coronavirus 2 Infection in |
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| Children. Journal of Pediatrics, 2022, 247, 22-28.e2. |

Fetal MRI compared with ultrasound for the diagnosis of obstructive genital malformations. Prenatal

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Inhibition of mitochondrial translation in fibroblasts from a patient expressing the KARS
67 p.(Pro228Leu) variant and presenting with sensorineural deafness, developmental delay, and lactic
acidosis. Human Mutation, 2018, 39, 2047-2059.
68 Central nervous system complications in adult cystinosis patients. Journal of Inherited Metabolic
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Disease, 2020, 43, 348-356.

Prenatal and postnatal presentations of corpus callosum agenesis with polymicrogyria caused by
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<i>EGP5</i> mutation. American Journal of Medical Genetics, Part A, 2017, 173, 706-711.
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70 Epileptic spasms in congenital disorders of glycosylation. Epileptic Disorders, 2017, 19, 15-23.

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The $\hat{€}$ œsalt and pepperâ€•pattern on renal ultrasound in a group of children with molecular-proven

Corpus callosum metrics predict severity of visuospatial and neuromotor dysfunctions in ARID1B
mutations with Coffinâ€"Siris syndrome. Psychiatric Genetics, 2019, 29, 237-242.
Focal Areas of High Signal Intensity in Children with Neurofibromatosis Type 1: Expected Evolution on MRI. American Journal of Neuroradiology, 2020, 41, 1733-1739.

Feasibility and Added Value of Fetal DTI Tractography in the Evaluation of an Isolated Short Corpus Callosum: Preliminary Results. American Journal of Neuroradiology, 2022, 43, 132-138.

A novel recurrent <i>LIS1</i> splice site mutation in classic lissencephaly. American Journal of Medical Genetics, Part A, 2017, 173, 561-564.

Biometric and morphological features on magnetic resonance imaging of fetal bladder in lower
95 urinary tract obstruction: new perspectives for fetal cystoscopy. Ultrasound in Obstetrics and Gynecology, 2020, 56, 86-95.

Cerebral blood flow and acute episodes of Leigh syndrome in neurometabolic disorders.
Developmental Medicine and Child Neurology, 2021, 63, 705-711.

Arterial Spin Labeling for the Etiological Workup of Intracerebral Hemorrhage in Children. Stroke,
2022, 53, 185-193.

Spectrum of Neuroradiologic Findings Associated with Monogenic Interferonopathies. American
Journal of Neuroradiology, 2022, 43, 2-10.

A neuropathological study of cerebrovascular abnormalities in a signal transducer and activator of
transcription 3ấ€"deficient patient. Journal of Allergy and Clinical Immunology, 2015, 136, 1418-1421.e5.

100 Arterial Spin Labeling and Central Precocious Puberty. Clinical Neuroradiology, 2020, 30, 137-144.
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Pediatric cardiac computed tomography angiography: Expert consensus from the Filiale de
101 Cardiologie PÃ@diatrique et CongÃ@nitale (FCPC) and the SociÃ ©tÃ@ FranÃ§aise dâ€ ${ }^{\top} M$ Imagerie Cardiaque et
101 Vasculaire diagnostique et interventionnelle (SFICV). Diagnostic and Interventional Imaging, 2020, 101,
335-345.
Deciphering the genetic and epigenetic landscape of pediatric bithalamic tumors. Brain Pathology, 2022, 32, el3039.

Prevalence of Venovenous Shunting and High-Output State Quantified with 4D Flow MRI in Patients with Fontan Circulation. Radiology: Cardiothoracic Imaging, 2021, 3, e210161.

Arterial spin labeling shows pre-epileptic tuber hyperperfusion in tuberous sclerosis complex. Neurology, 2016, 86, 1744-1745.

Neonatal factors related to survival and intellectual and developmental outcome of patients with early-onset urea cycle disorders. Molecular Genetics and Metabolism, 2020, 130, 110-117.
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Variants in the MIPEP gene presenting with complex neurological phenotype without cardiomyopathy,
107 impair OXPHOS protein maturation and lead to a reduced OXPHOS abundance in patient cells. Molecular Genetics and Metabolism, 2021, 134, 267-273.
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The genomic landscape of dysembryoplastic neuroepithelial tumours and a comprehensive analysis of recurrent cases. Neuropathology and Applied Neurobiology, 2022, 48, .

| 111 | Arterial abnormalities identified in kidneys transplanted into children during the COVID-19 pandemic. American Journal of Transplantation, 2021, 21, 1937-1943. | 2.6 | 3 |
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| 112 | Pineal alveolar rhabdomyosarcoma with PAX3:NCOA2 fusion inducing OLIG2 expression, a potential pitfall in the central nervous system. Histopathology, 2021, 79, 437-439. | 1.6 | 3 |
| 113 | A novel case of cribriform neuroepithelial tumor: A potential diagnostic pitfall in the ventricular system. Pediatric Blood and Cancer, 2021, 68, e29037. | 0.8 | 3 |
| 114 | Brain perfusion magnetic resonance imaging using pseudocontinuous arterial spin labeling in 314 dogs and cats. Journal of Veterinary Internal Medicine, 2021, 35, 2327-2341. | 0.6 | 3 |
| 115 | A novel LARGE1-AFF2 fusion expanding the molecular alterations associated with the methylation class of neuroepithelial tumors with PATZ1 fusions. Acta Neuropathologica Communications, 2022, 10, 15. | 2.4 | 3 |

119 Imaging features of complete congenital atresia of left coronary artery. Diagnostic and Interventional Imaging, 2020, 101, 421-423.
1.8 ..... 2
120 Neural basis of interindividual variability in social perception in typically developing children and1.6adolescents using diffusion tensor imaging. Scientific Reports, 2020, 10, 6379.

Acute surgical management of children with ruptured brain arteriovenous malformation. Journal of Neurosurgery: Pediatrics, 2021, 27, 437-445.
$0.8 \quad 2$

Hydrocephalus in children with ruptured cerebral arteriovenous malformation. Journal of Neurosurgery: Pediatrics, 2020, 26, 283-287.

Aortic atresia and interrupted aortic arch communicating through external carotid anastomosis. Cardiology in the Young, 2019, 29, 699-700.

