

# Roelof-Jan Oostra

## List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

91 papers	1,724 citations	22 h-index	38 g-index
98 ext. papers	2,005 ext. citations	4 avg, IF	4.38 L-index

#	Paper	IF	Citations
91	Primary pathogenic mtDNA mutations in multigeneration pedigrees with Leber hereditary optic neuropathy. <i>American Journal of Human Genetics</i> , <b>1996</b> , 59, 481-5	11	221
90	An interactive three-dimensional digital atlas and quantitative database of human development. <i>Science</i> , <b>2016</b> , 354,	33.3	98
89	Sequence analysis of the mitochondrial genomes from Dutch pedigrees with Leber hereditary optic neuropathy. <i>American Journal of Human Genetics</i> , <b>2003</b> , 72, 1460-9	11	98
88	Lessons from BWS twins: complex maternal and paternal hypomethylation and a common source of haematopoietic stem cells. <i>European Journal of Human Genetics</i> , <b>2009</b> , 17, 1625-34	5.3	85
87	The anatomy in relation to injury of the lateral collateral ligaments of the ankle: a current concepts review. <i>Clinical Anatomy</i> , <b>2008</b> , 21, 619-26	2.5	83
86	Leber's hereditary optic neuropathy: correlations between mitochondrial genotype and visual outcome. <i>Journal of Medical Genetics</i> , <b>1994</b> , 31, 280-6	5.8	76
85	A gene for X-linked optic atrophy is closely linked to the Xp11.4-Xp11.2 region of the X chromosome. <i>American Journal of Human Genetics</i> , <b>1997</b> , 61, 934-9	11	52
84	Pectus excavatum and carinatum. <i>European Journal of Medical Genetics</i> , <b>2014</b> , 57, 414-7	2.6	46
83	Congenital anomalies in the teratological collection of Museum Vrolik in Amsterdam, The Netherlands. II: Skeletal dysplasias. <i>American Journal of Medical Genetics Part A</i> , <b>1998</b> , 77, 116-134		43
82	Blomstrand osteochondrodysplasia: three novel cases and histological evidence for heterogeneity. <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , <b>2000</b> , 436, 28-35	5.1	41
81	Development of a mRNA profiling multiplex for the inference of organ tissues. <i>International Journal of Legal Medicine</i> , <b>2013</b> , 127, 891-900	3.1	38
80	Development of the hearts of lizards and snakes and perspectives to cardiac evolution. <i>PLoS ONE</i> , <b>2013</b> , 8, e63651	3.7	37
79	The mitochondrial DNA mutation ND6*14,484C associated with leber hereditary optic neuropathy, leads to deficiency of complex I of the respiratory chain. <i>Biochemical and Biophysical Research Communications</i> , <b>1995</b> , 215, 1001-5	3.4	37
78	Leber's hereditary optic neuropathy: no significant evidence for primary or secondary pathogenicity of the 15257 mutation. <i>Human Genetics</i> , <b>1994</b> , 94, 265-70	6.3	35
77	Congenital anomalies in the teratological collection of Museum Vrolik in Amsterdam, The Netherlands. I: Syndromes with multiple congenital anomalies. <i>American Journal of Medical Genetics Part A</i> , <b>1998</b> , 77, 100-115		34
76	In patients with pseudoxanthoma elasticum a thicker and more elastic carotid artery is associated with elastin fragmentation and proteoglycans accumulation. <i>Ultrasound in Medicine and Biology</i> , <b>2004</b> , 30, 1041-8	3.5	34
75	No evidence for 'skewed' inactivation of the X-chromosome as cause of Leber's hereditary optic neuropathy in female carriers. <i>Human Genetics</i> , <b>1996</b> , 97, 500-5	6.3	31

74	The hypertrabeculated (noncompacted) left ventricle is different from the ventricle of embryos and ectothermic vertebrates. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , <b>2016</b> , 1863, 1696-706	4.9	29
73	Mutations in the T (brachyury) gene cause a novel syndrome consisting of sacral agenesis, abnormal ossification of the vertebral bodies and a persistent notochordal canal. <i>Journal of Medical Genetics</i> , <b>2014</b> , 51, 90-7	5.8	28
72	Brachmann-de Lange syndrome "avant la lettre". <i>American Journal of Medical Genetics Part A</i> , <b>1994</b> , 52, 267-8		28
71	The geometrical precision of virtual bone models derived from clinical computed tomography data for forensic anthropology. <i>International Journal of Legal Medicine</i> , <b>2017</b> , 131, 1155-1163	3.1	27
70	Towards a 3-dimensional atlas of the developing human embryo: the Amsterdam experience. <i>Reproductive Toxicology</i> , <b>2012</b> , 34, 225-36	3.4	25
69	Congenital anomalies in the teratological collection of museum Vrolik in Amsterdam, the Netherlands. v: conjoined and acardiac twins. <i>American Journal of Medical Genetics Part A</i> , <b>1998</b> , 80, 74-89		21
68	Historical aspects of the study of malformations in The Netherlands. <i>American Journal of Medical Genetics Part A</i> , <b>1998</b> , 77, 91-99		20
67	Smith-Lemli-Opitz syndrome diagnosed in a 130-year-old anatomical specimen. <i>American Journal of Medical Genetics Part A</i> , <b>1997</b> , 68, 257-9		18
66	The development of the human notochord. <i>PLoS ONE</i> , <b>2018</b> , 13, e0205752	3.7	18
65	A review of the embryological development and associated developmental abnormalities of the sternum in the light of a rare palaeopathological case of sternal clefting. <i>HOMO- Journal of Comparative Human Biology</i> , <b>2013</b> , 64, 129-41	0.5	16
64	Severe acrofacial dysostosis with orofacial clefting and tetraphocomelia diagnosed in the plaster cast of a 100-year-old anatomical specimen. <i>American Journal of Medical Genetics Part A</i> , <b>1998</b> , 78, 195-7		16
63	Malformations of the axial skeleton in Museum Vrolik I: homeotic transformations and numerical anomalies. <i>American Journal of Medical Genetics, Part A</i> , <b>2005</b> , 134, 268-81	2.5	16
62	Leber's hereditary optic neuropathy: implications of the sex ratio for linkage studies in families with the 3460 ND1 mutation. <i>Eye</i> , <b>1995</b> , 9 ( Pt 4), 513-6	4.4	16
61	Congenital anomalies in the teratological collection of Museum Vrolik in Amsterdam, The Netherlands. III: primary field defects, sequences, and other complex anomalies. <i>American Journal of Medical Genetics Part A</i> , <b>1998</b> , 80, 46-59		15
60	Malformations of the axial skeleton in the museum Vrolik: II: craniosynostoses and suture-related conditions. <i>American Journal of Medical Genetics, Part A</i> , <b>2005</b> , 136A, 327-42	2.5	15
59	Study on the three-dimensional orientation of the posterior facet of the subtalar joint using simulated weight-bearing CT. <i>Journal of Orthopaedic Research</i> , <b>2019</b> , 37, 197-204	3.8	15
58	Virtual forensic anthropology: The accuracy of osteometric analysis of 3D bone models derived from clinical computed tomography (CT) scans. <i>Forensic Science International</i> , <b>2019</b> , 304, 109963	2.6	14
57	The accuracy of 3D virtual bone models of the pelvis for morphological sex estimation. <i>International Journal of Legal Medicine</i> , <b>2019</b> , 133, 1853-1860	3.1	14

56	Dutch population specific sex estimation formulae using the proximal femur. <i>Forensic Science International</i> , <b>2018</b> , 286, 268.e1-268.e8	2.6	14
55	Postmortem imaging exposed: an aid in MR imaging of musculoskeletal structures. <i>Skeletal Radiology</i> , <b>2013</b> , 42, 467-72	2.7	13
54	Congenital anomalies in the teratological collection of Museum Vrolik in Amsterdam, The Netherlands. IV: closure defects of the neural tube. <i>American Journal of Medical Genetics Part A</i> , <b>1998</b> , 80, 60-73		13
53	Frederik Ruysch (1638-1731): Historical perspective and contemporary analysis of his teratological legacy. <i>American Journal of Medical Genetics, Part A</i> , <b>2017</b> , 173, 16-41	2.5	12
52	Estimation of the postmortem interval based on the human decomposition process. <i>Journal of Clinical Forensic and Legal Medicine</i> , <b>2019</b> , 61, 122-127	1.7	11
51	Colourimetric analysis of thermally altered human bone samples. <i>Scientific Reports</i> , <b>2019</b> , 9, 8923	4.9	10
50	Trapezius aplasia: indications for a dual developmental origin of the trapezius muscle. <i>Clinical Anatomy</i> , <b>2006</b> , 19, 547-9	2.5	10
49	Homozygous DMRT2 variant associates with severe rib malformations in a newborn. <i>American Journal of Medical Genetics, Part A</i> , <b>2018</b> , 176, 1216-1221	2.5	9
48	Temperature estimations of heated bone: A questionnaire-based study of accuracy and precision of interpretation of bone colour by forensic and physical anthropologists. <i>Legal Medicine</i> , <b>2017</b> , 29, 22-28	1.9	9
47	Thanatophoric dysplasia type II with encephalocele and aortic hypoplasia diagnosed in an anatomical specimen. <i>American Journal of Medical Genetics Part A</i> , <b>2003</b> , 118A, 64-7		9
46	A 100-year-old anatomical specimen presenting with boomerang-like skeletal dysplasia: diagnostic strategies and outcome. <i>American Journal of Medical Genetics Part A</i> , <b>1999</b> , 85, 134-9		9
45	Hitherto unknown detailed muscle anatomy in an 8-week-old embryo. <i>Journal of Anatomy</i> , <b>2018</b> , 233, 243-254	2.9	8
44	Luminescence of thermally altered human skeletal remains. <i>International Journal of Legal Medicine</i> , <b>2017</b> , 131, 1165-1177	3.1	8
43	On the many faces of Leber hereditary optic neuropathy. <i>Clinical Genetics</i> , <b>1997</b> , 51, 388-93	4	8
42	Bifid ribs and unusual vertebral anomalies diagnosed in an anatomical specimen. Gorlin syndrome?. <i>American Journal of Medical Genetics, Part A</i> , <b>2006</b> , 140, 2135-8	2.5	8
41	Reply to Hofmann et al. <i>American Journal of Human Genetics</i> , <b>1998</b> , 62, 492-5	11	7
40	Respiratory chain function in Leber's hereditary optic neuropathy: lack of correlation with clinical disease. <i>Journal of Inherited Metabolic Disease</i> , <b>1993</b> , 16, 531-3	5.4	7
39	Simultaneous occurrence of the 11778 (ND4) and the 9438 (COX III) mtDNA mutations in Leber hereditary optic neuropathy: molecular, biochemical, and clinical findings. <i>American Journal of Human Genetics</i> , <b>1995</b> , 57, 954-7	11	7

38	The effect of repeated freeze-thaw cycles on human muscle tissue visualized by postmortem computed tomography (PMCT). <i>Clinical Anatomy</i> , <b>2017</b> , 30, 799-804	2.5	6
37	Sinus venosus incorporation: contentious issues and operational criteria for developmental and evolutionary studies. <i>Journal of Anatomy</i> , <b>2019</b> , 234, 583-591	2.9	6
36	GATA6 mutations: Characterization of two novel patients and a comprehensive overview of the GATA6 genotypic and phenotypic spectrum. <i>American Journal of Medical Genetics, Part A</i> , <b>2019</b> , 179, 1836-1845	2.5	6
35	Single-site neural tube closure in human embryos revisited. <i>Clinical Anatomy</i> , <b>2017</b> , 30, 988-999	2.5	6
34	Abnormal growth of the proximal femur due to apophyseal-epiphyseal coalescence resulting in coxa valga—a report of two cases in adolescents. <i>Monthly Notices of the Royal Astronomical Society: Letters</i> , <b>2011</b> , 82, 507-9	4.3	6
33	Mitochondrial DNA analysis as a diagnostic tool in singleton cases of Leber's hereditary optic neuropathy. <i>Ophthalmic Paediatrics and Genetics</i> , <b>1993</b> , 14, 109-15		6
32	The applicability of forensic time since death estimation methods for buried bodies in advanced decomposition stages. <i>PLoS ONE</i> , <b>2020</b> , 15, e0243395	3.7	6
31	Single origin of the epithelium of the human middle ear. <i>Mechanisms of Development</i> , <b>2019</b> , 158, 103556	1.7	5
30	Evaluation of collimated polarized light imaging for real-time intraoperative selective nerve identification in the human hand. <i>Biomedical Optics Express</i> , <b>2017</b> , 8, 4122-4134	3.5	5
29	History and highlights of the teratological collection in the Museum Anatomicum of Leiden University, The Netherlands. <i>American Journal of Medical Genetics, Part A</i> , <b>2018</b> , 176, 618-637	2.5	4
28	Absence of the spleen(s) in conjoined twins: a diagnostic clue of laterality defects? Radiological study of historical specimens. <i>Pediatric Radiology</i> , <b>2012</b> , 42, 653-9	2.8	4
27	Dutch teratological collections and their artistic portrayals. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , <b>2021</b> , 187, 283-295	3.1	4
26	When the right (Drug) should be left: Prenatal drug exposure and heterotaxy syndrome. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , <b>2016</b> , 106, 573-9		4
25	Congenital anomalies in the teratological collection of Museum Vrolik in Amsterdam, The Netherlands. V: conjoined and acardiac twins. <i>American Journal of Medical Genetics Part A</i> , <b>1998</b> , 80, 74-89		4
24	The dimensions of the tarsal sinus and canal in different foot positions and its clinical implications. <i>Clinical Anatomy</i> , <b>2017</b> , 30, 1049-1057	2.5	3
23	Paleodysmorphology and paleoteratology: Diagnosing and interpreting congenital conditions of the skeleton in anthropological contexts. <i>Clinical Anatomy</i> , <b>2016</b> , 29, 878-91	2.5	3
22	Individualised and non-contact post-mortem interval determination of human bodies using visible and thermal 3D imaging. <i>Nature Communications</i> , <b>2021</b> , 12, 5997	17.4	3
21	The distal radial physis: Exploring normal anatomy on MRI enables interpretation of stress related changes in young gymnasts. <i>European Journal of Sport Science</i> , <b>2020</b> , 20, 1197-1205	3.9	3

20	Amsterdam Research Initiative for Sub-surface Taphonomy and Anthropology (ARISTA) - A taphonomic research facility in the Netherlands for the study of human remains. <i>Forensic Science International</i> , <b>2020</b> , 317, 110483	2.6	3
19	Characterizing the coalescence area of conjoined twins to elucidate congenital disorders in singletons. <i>Clinical Anatomy</i> , <b>2021</b> , 34, 845-858	2.5	3
18	Comment on 'A DNA extraction method for small quantities of bone for high-quality genotyping' by Caputo et al. <i>Forensic Science International: Genetics</i> , <b>2019</b> , 40, e243-e244	4.3	2
17	Detection of G1138A Mutation of the FGFR3 Gene in Tooth Material from a 180-Year-Old Museological Achondroplastic Skeleton. <i>Genes</i> , <b>2017</b> , 8,	4.2	2
16	Segmentation anomalies of vertebrae and ribs with other abnormalities of blastogenesis: syndromes or associations?. <i>Fetal and Pediatric Pathology</i> , <b>2005</b> , 24, 331-45	1.7	2
15	The triangular fibrocartilage complex on high-resolution 3T MRI in healthy adolescents: the thin line between asymptomatic findings and pathology. <i>Skeletal Radiology</i> , <b>2021</b> , 50, 2195-2204	2.7	2
14	Prenatal Evidence of Persistent Notochord and Absent Sacrum Caused by a Mutation in the T (Brachyury) Gene. <i>Case Reports in Obstetrics and Gynecology</i> , <b>2016</b> , 2016, 7625341	0.8	2
13	Development pattern of tracheal cartilage in human embryos. <i>Clinical Anatomy</i> , <b>2021</b> , 34, 668-672	2.5	2
12	Congenital anomalies in the teratological collection of Museum Vrolik in Amsterdam, The Netherlands. II: Skeletal dysplasias. <i>American Journal of Medical Genetics Part A</i> , <b>1998</b> , 77, 116-34		2
11	The Pronephros; a Fresh Perspective. <i>Integrative and Comparative Biology</i> , <b>2019</b> , 59, 29-47	2.8	1
10	No evidence for skewed inactivation of the X-chromosome as cause of Leber's hereditary optic neuropathy in female carriers <b>1996</b> , 97, 500		1
9	Bilateral symmetry of the subtalar joint facets and the relationship between the morphology and osteoarthritic changes. <i>Clinical Anatomy</i> , <b>2020</b> , 33, 997-1006	2.5	1
8	Phosphorescence of thermally altered human bone. <i>International Journal of Legal Medicine</i> , <b>2021</b> , 135, 1025-1034	3.1	1
7	Higher spatial resolution improves the interpretation of the extent of ventricular trabeculation. <i>Journal of Anatomy</i> , <b>2021</b> ,	2.9	1
6	Congenital anomalies in the teratological collection of Museum Vrolik in Amsterdam, The Netherlands. I: Syndromes with multiple congenital anomalies. <i>American Journal of Medical Genetics Part A</i> , <b>1998</b> , 77, 100-15		1
5	Gunshot trauma in human long bones: towards practical diagnostic guidance for forensic anthropologists.. <i>Forensic Science, Medicine, and Pathology</i> , <b>2022</b> , 1	1.5	0
4	Unilateral partial absence of the fallopian tube in Williams syndrome: a new feature?. <i>Clinical Dysmorphology</i> , <b>2007</b> , 16, 195-196	0.9	
3	Midline crossing pulmonary vein: right upper lobe dual venous drainage, with partial anomalous venous return of the right lung into a persistent left superior vena cava. <i>Surgical and Radiologic Anatomy</i> , <b>2021</b> , 1	1.4	

2      3D Atlas of the developing human. *FASEB Journal*, **2012**, 26, 530.6      0.9

1      Mechanical or thermal damage: differentiating between underlying mechanisms as a cause of bone fractures.. *International Journal of Legal Medicine*, **2022**, 1      3.1