Francois Vialard

List of Publications by Year in Descending Order

139 papers	2,798 citations	28 h-index	43 g-index
166 ext. papers	3,324 ext. citations	3.2 avg, IF	4.56 L-index

#	Paper	IF	Citations
139	The Eutopic Endometrium Proteome in Endometriosis Reveals Candidate Markers and Molecular Mechanisms of Physiopathology.. *Diagnostics*, **2022**, 12,	3.8	1
138	Azoospermia and reciprocal translocations: should whole-exome sequencing be recommended?. *Basic and Clinical Andrology*, **2021**, 31, 27	2.8	0
137	Effects of the implementation of second-line prenatal cell-free DNA testing on termination of pregnancy in a French perinatal network. *European Journal of Obstetrics, Gynecology and Reproductive Biology*, **2021**, 267, 36-41	2.4	
136	Human testis-expressed (TEX) genes: a review focused on spermatogenesis and male fertility. *Basic and Clinical Andrology*, **2021**, 31, 9	2.8	2
135	Preimplantation factor modulates trophoblastic invasion throughout the decidualization of human endometrial stromal cells. *Reproductive Biology and Endocrinology*, **2021**, 19, 96	5	1
134	Metabolic Diseases and Down Syndrome: How Are They Linked Together?. *Biomedicines*, **2021**, 9,	4.8	5
133	The Mare: A Pertinent Model for Human Assisted Reproductive Technologies?. *Animals*, **2021**, 11,	3.1	3
132	STAG3 homozygous missense variant causes primary ovarian insufficiency and male non-obstructive azoospermia. *Molecular Human Reproduction*, **2020**, 26, 665-677	4.4	11
131	A high level of tetrasomy 9p mosaicism but no clinical manifestations other than moderate oligozoospermia with chromosomally balanced sperm: a case report. *Journal of Assisted Reproduction and Genetics*, **2020**, 37, 573-577	3.4	1
130	Prenatal diagnosis of 2q13 duplications: The crucial role of the family survey in genetic counseling on novel copy number variations. *European Journal of Medical Genetics*, **2020**, 63, 103956	2.6	0
129	Polymorphisms and endometriosis: a systematic review and meta-analyses. *Human Reproduction Update*, **2020**, 26, 73-102	15.8	24
128	COVID-19 and Down's syndrome: are we heading for a disaster?. *European Journal of Human Genetics*, **2020**, 28, 1477-1478	5.3	6
127	Prenatal findings in 1p36 deletion syndrome: New cases and a literature review. *Prenatal Diagnosis*, **2019**, 39, 871-882	3.2	7
126	Down syndrome and infertility: what support should we provide?. *Journal of Assisted Reproduction and Genetics*, **2019**, 36, 1063-1067	3.4	9
125	Genetic defects in human azoospermia. *Basic and Clinical Andrology*, **2019**, 29, 4	2.8	18
124	Chromosomal microarray analysis in fetuses with an isolated congenital heart defect: A retrospective, nationwide, multicenter study in France. *Prenatal Diagnosis*, **2019**, 39, 464-470	3.2	9
123	Maternal obesity influences expression and DNA methylation of the adiponectin and leptin systems in human third-trimester placenta. *Clinical Epigenetics*, **2019**, 11, 20	7.7	39

122	A MEI1 homozygous missense mutation associated with meiotic arrest in a consanguineous family. *Human Reproduction*, **2018**, 33, 1034-1037	5.7	27
121	Are leptin and adiponectin involved in recurrent pregnancy loss?. *Journal of Obstetrics and Gynaecology Research*, **2018**, 44, 1015-1022	1.9	8
120	Diagnostic yield of chromosomal microarray analysis in fetuses with isolated increased nuchal translucency: a French multicenter study. *Ultrasound in Obstetrics and Gynecology*, **2018**, 52, 715-721	5.8	25
119	Adiponectin regulates glycogen metabolism at the human fetal–maternal interface. *Journal of Molecular Endocrinology*, **2018**, 61, 139-152	4.5	9
118	First prenatal diagnosis of a 'pure' 9q34.3 deletion (Kleefstra syndrome): A case report and literature review. *Journal of Obstetrics and Gynaecology Research*, **2018**, 44, 570-575	1.9	3
117	Testicular Spermatozoa Are of Better Quality Than Epididymal Spermatozoa in Patients With Obstructive Azoospermia. *Urology*, **2017**, 103, 106-111	1.6	17
116	The value of cytogenetic analysis of the product of conception before preimplantation genetic screening. *Human Reproduction*, **2017**, 32, 477-478	5.7	
115	Is anagrelide safe during pregnancy?. *Journal of Gynecology Obstetrics and Human Reproduction*, **2017**, 46, 697-699	1.9	2
114	Adiponectin limits differentiation and trophoblast invasion in human endometrial cells. *Journal of Molecular Endocrinology*, **2017**, 59, 285-297	4.5	5
113	Placental perfusion: interest and limits. *Journal of Maternal-Fetal and Neonatal Medicine*, **2017**, 30, 1347-1348	1	5
112	PreImplantation Factor and Endocrinology of Implantation and Establishment of Early Pregnancy: A Contemporary View. *Pediatric Endocrinology Reviews*, **2017**, 15, 147-158	1.1	3
111	The emerging microduplication 3q13.31: Expanding the genotype-phenotype correlations of the reciprocal microdeletion 3q13.31 syndrome. *European Journal of Medical Genetics*, **2016**, 59, 463-9	2.6	5
110	Is intracouple assisted reproductive technology an option for men with large-headed spermatozoa? A literature review and a decision guide proposal. *Basic and Clinical Andrology*, **2016**, 26, 8	2.8	3
109	To be or not to be [fertile], that is the question. *Basic and Clinical Andrology*, **2016**, 26, 12	2.8	2
108	PreImplantation Factor (PIF*) endogenously prevents preeclampsia: Promotes trophoblast invasion and reduces oxidative stress. *Journal of Reproductive Immunology*, **2016**, 114, 58-64	4.2	16
107	A French multicenter study of over 700 patients with 22q11 deletions diagnosed using FISH or aCGH. *European Journal of Human Genetics*, **2016**, 24, 844-51	5.3	28
106	Could Digital PCR Be an Alternative as a Non-Invasive Prenatal Test for Trisomy 21: A Proof of Concept Study. *PLoS ONE*, **2016**, 11, e0155009	3.7	26
105	Pregnancy outcomes in prenatally diagnosed 47, XXX and 47, XYY syndromes: a 30-year French, retrospective, multicentre study. *Prenatal Diagnosis*, **2016**, 36, 523-9	3.2	10

104	Preimplantation factor is an anti-apoptotic effector in human trophoblasts involving p53 signaling pathway. *Cell Death and Disease*, **2016**, 7, e2504	9.8	19
103	Adiponectin Inhibits Nutrient Transporters and Promotes Apoptosis in Human Villous Cytotrophoblasts: Involvement in the Control of Fetal Growth. *Biology of Reproduction*, **2016**, 94, 111	3.9	24
102	Use of anticancer agents in gynecological oncology during pregnancy: a systematic review of maternal pharmacokinetics and transplacental transfer. *Expert Opinion on Drug Metabolism and Toxicology*, **2016**, 12, 523-31	5.5	7
101	Aneuploidy: the impact of chromosome imbalance on nuclear organization and overall genome expression. *Clinical Genetics*, **2016**, 90, 35-48	4	9
100	Trophoblast syncytialisation necessitates mitochondrial function through estrogen-related receptor-□activation. *Molecular Human Reproduction*, **2015**, 21, 206-16	4.4	25
99	Low-level mosaicism of a de novo derivative chromosome 9 from a t(5;9)(q35.1;q34.3) has a major phenotypic impact. *European Journal of Medical Genetics*, **2015**, 58, 346-50	2.6	5
98	The roles of leptin and adiponectin at the fetal-maternal interface in humans. *Hormone Molecular Biology and Clinical Investigation*, **2015**, 24, 47-63	1.3	21
97	Prenatal diagnosis of 24 cases of microduplication 22q11.2: an investigation of phenotype-genotype correlations. *Prenatal Diagnosis*, **2015**, 35, 35-43	3.2	17
96	Are de novo rea(21;21) chromosomes really de novo?. *Clinical Case Reports (discontinued)*, **2015**, 3, 786-9	0.7	4
95	Prevalence of recurrent pathogenic microdeletions and microduplications in over 9500 pregnancies. *Prenatal Diagnosis*, **2015**, 35, 801-9	3.2	191
94	Involvement of estrogen-related receptor-□and mitochondrial content in intrauterine growth restriction and preeclampsia. *Fertility and Sterility*, **2015**, 104, 483-90	4.8	37
93	Non-invasive prenatal testing for trisomy 21 based on analysis of cell-free fetal DNA circulating in the maternal plasma. *Prenatal Diagnosis*, **2015**, 35, 471-6	3.2	12
92	A human morphologically normal spermatozoon may have noncondensed chromatin. *Andrologia*, **2015**, 47, 879-86	2.4	9
91	BACs-on-Beads□(BoBs)□assay for the genetic evaluation of prenatal samples and products of conception. *Methods in Molecular Biology*, **2015**, 1227, 259-78	1.4	8
90	RHOXF2 gene, a new candidate gene for spermatogenesis failure. *Basic and Clinical Andrology*, **2014**, 24, 3	2.8	4
89	A French collaborative survey of 272 fetuses with 22q11.2 deletion: ultrasound findings, fetal autopsies and pregnancy outcomes. *Prenatal Diagnosis*, **2014**, 34, 424-30	3.2	38
88	High-magnification selection of spermatozoa prior to oocyte injection: confirmed and potential indications. *Reproductive BioMedicine Online*, **2014**, 28, 6-13	4	39
87	Prenatal phenotype of Williams-Beuren syndrome and of the reciprocal duplication syndrome. *Clinical Case Reports (discontinued)*, **2014**, 2, 25-32	0.7	12

86	Molecular characterization of 39 de novo sSMC: contribution to prognosis and genetic counselling, a prospective study. *Clinical Genetics*, **2014**, 85, 233-44	4	21
85	Preimplantation factor (PIF) promotes human trophoblast invasion. *Biology of Reproduction*, **2014**, 91, 118	3.9	28
84	Chronic excessive alcohol consumption and male fertility: a case report on reversible azoospermia and a literature review. *Alcohol and Alcoholism*, **2014**, 49, 42-4	3.5	24
83	Pregnancy outcomes of prenatally diagnosed Turner syndrome: a French multicenter retrospective study including a series of 975 cases. *Prenatal Diagnosis*, **2014**, 34, 1133-8	3.2	17
82	Birth of a boy with isolated short stature after prenatal diagnosis of a Xp22.3 nullosomy due to an inherited t(X;15) (p22.3;p10) translocation. *Clinical Case Reports (discontinued)*, **2014**, 2, 98-102	0.7	1
81	Sperm FISH analysis of a 44,X,der(Y),t(Y;15)(q12;q10)pat,rob(13;14)(q10;q10)mat complex chromosome rearrangement. *Andrologia*, **2014**, 46, 576-82	2.4	3
80	Associations between Individual and Combined Polymorphisms of the TNF and VEGF Genes and the Embryo Implantation Rate in Patients Undergoing In Vitro Fertilization (IVF) Programs. *PLoS ONE*, **2014**, 9, e108287	3.7	18
79	Selection of normal spermatozoa with a vacuole-free head (x6300) improves selection of spermatozoa with intact DNA in patients with high sperm DNA fragmentation rates. *Andrologia*, **2013**, 45, 163-70	2.4	49
78	High-magnification sperm selection does not decrease the aneuploidy rate in patients who are heterozygous for reciprocal translocations. *Journal of Assisted Reproduction and Genetics*, **2013**, 30, 525-30	3.4	10
77	A genome-wide DNA methylation study in azoospermia. *Andrology*, **2013**, 1, 815-21	4.2	26
76	Small human sperm vacuoles observed under high magnification are pocket-like nuclear concavities linked to chromatin condensation failure. *Reproductive BioMedicine Online*, **2013**, 27, 201-11	4	29
75	Tumor necrosis factor-308 polymorphism increases the embryo implantation rate in women undergoing in vitro fertilization. *Human Reproduction*, **2013**, 28, 2774-83	5.7	8
74	The nature of human sperm head vacuoles: a systematic literature review. *Basic and Clinical Andrology*, **2013**, 23, 3	2.8	14
73	Application of a new molecular technique for the genetic evaluation of products of conception. *Prenatal Diagnosis*, **2013**, 33, 32-41	3.2	14
72	Contraindication of ART following a sperm FISH analysis, even though only 12% of the spermatozoa had enlarged heads. *Systems Biology in Reproductive Medicine*, **2013**, 59, 214-7	2.9	3
71	DNA fragmentation is higher in spermatozoa with chromosomally unbalanced content in men with a structural chromosomal rearrangement. *Andrology*, **2013**, 1, 632-8	4.2	18
70	Can one translocation impact the meiotic segregation of another translocation? A sperm-FISH analysis of a 46,XY,t(1;16)(q21;p11.2),t(8;9) (q24.3;p24) patient and his 46,XY,t(8;9)(q24.3;p24) brother and cousin. *Molecular Human Reproduction*, **2013**, 19, 109-17	4.4	4
69	Follicular fluid protein content (FSH, LH, PG4, E2 and AMH) and polar body aneuploidy. *Journal of Assisted Reproduction and Genetics*, **2012**, 29, 1123-34	3.4	8

68	The vascular endothelial growth factor (VEGF) +405´G/C polymorphism and its relationship with recurrent implantation failure in women in an IVF programme with ICSI. *Journal of Assisted Reproduction and Genetics*, **2012**, 29, 1415-20	3.4	18
67	Sperm FISH analysis of a 46,XY,t(3;6)(p24;p21.2),inv (8)(p11;2q21.2) double chromosomal rearrangement. *Reproductive BioMedicine Online*, **2012**, 24, 219-23	4	6
66	The high frequency of sperm aneuploidy in klinefelter patients and in nonobstructive azoospermia is due to meiotic errors in euploid spermatocytes. *Journal of Andrology*, **2012**, 33, 1352-9		29
65	Cryopreservation of human spermatozoa decreases the number of motile normal spermatozoa, induces nuclear vacuolization and chromatin decondensation. *Journal of Andrology*, **2012**, 33, 1371-8		39
64	Prenatal diagnosis of the duplication 17p11.2 associated with Potocki-Lupski syndrome in a foetus presenting with mildly dysmorphic features. *European Journal of Medical Genetics*, **2012**, 55, 723-6	2.6	9
63	Prenatal BACs-on-Beads□the prospective experience of five prenatal diagnosis laboratories. *Prenatal Diagnosis*, **2012**, 32, 329-35	3.2	37
62	Identification of a new recurrent aurora kinase C mutation in both European and African men with macrozoospermia. *Human Reproduction*, **2012**, 27, 3337-46	5.7	40
61	MLPA and sequence analysis of DPY19L2 reveals point mutations causing globozoospermia. *Human Reproduction*, **2012**, 27, 2549-58	5.7	48
60	Genetic polymorphisms influence the ovarian response to rFSH stimulation in patients undergoing in vitro fertilization programs with ICSI. *PLoS ONE*, **2012**, 7, e38700	3.7	53
59	Predisposition to aneuploidy in the oocyte. *Cytogenetic and Genome Research*, **2011**, 133, 127-35	1.9	29
58	The chromosomal risk in sperm from heterozygous Robertsonian translocation carriers is related to´the´sperm count and the translocation type. *Fertility and Sterility*, **2011**, 96, 1337-43	4.8	21
57	Inverse correlation between chromatin condensation and sperm head size in a case of enlarged sperm heads. *Reproductive BioMedicine Online*, **2011**, 23, 711-6	4	10
56	Williams-Beuren syndrome: the prenatal phenotype. *American Journal of Obstetrics and Gynecology*, **2011**, 205, e6-8	6.4	21
55	A recurrent deletion of DPY19L2 causes infertility in man by blocking sperm head elongation and acrosome formation. *American Journal of Human Genetics*, **2011**, 88, 351-61	11	133
54	Apport de l□exploration cytogh□tique et ultrastructurale dans le pronostic de fertilit□des sujets globozoospermiques. *Basic and Clinical Andrology*, **2011**, 21, 240-246	2.8	4
53	De la nature des vacuoles spermatiques aux r□sultats et indications de l□MSI (intracytoplasmic morphologically selected sperm injection). *Basic and Clinical Andrology*, **2011**, 21, 234-239	2.8	3
52	Avons-nous perc□le myst□e de la globozoospermie ?. *Basic and Clinical Andrology*, **2011**, 21, 230-233	2.8	1
51	Prenatal BACs-on-Beads□ a new technology for rapid detection of aneuploidies and microdeletions in prenatal diagnosis. *Prenatal Diagnosis*, **2011**, 31, 500-8	3.2	39

50	Large human sperm vacuoles observed in motile spermatozoa under high magnification: nuclear thumbprints linked to failure of chromatin condensation. *Human Reproduction*, **2011**, 26, 1650-8	5.7	101
49	Pregnancy outcomes in 188 French cases of prenatally diagnosed Klinefelter syndrome. *Human Reproduction*, **2011**, 26, 2570-5	5.7	22
48	Gh†tique et infertilit˜masculine **2011**, 359-369		
47	Analyse du premier globule polaire et diagnostic pr˜conceptionnel: †ats des lieux **2011**, 389-399		
46	Can intracytoplasmic morphologically selected sperm injection be used to select normal-sized sperm heads in infertile patients with macrocephalic sperm head syndrome?. *Fertility and Sterility*, **2010**, 93, 1347.e1-5	4.8	27
45	Une alt†ation gh†tique peut-elle ‰re ˆ l'origine d'une infertilit˜masculine et f˜minine ?. *Basic and Clinical Andrology*, **2010**, 20, 120-122	2.8	
44	Impact of freezing/thawing technique on sperm DNA integrity in HIV-1 patients. *Journal of Assisted Reproduction and Genetics*, **2010**, 27, 415-21	3.4	14
43	Are zona pellucida laser drilling and polar body biopsy safe for in vitro matured oocytes?. *Journal of Assisted Reproduction and Genetics*, **2010**, 27, 423-7	3.4	15
42	The Aurora Kinase C c.144delC mutation causes meiosis I arrest in men and is frequent in the North African population. *Human Molecular Genetics*, **2009**, 18, 1301-9	5.6	77
41	Preconceptional diagnosis for Robertsonian translocation as an alternative to preimplantation genetic diagnosis in two situations: a pilot study. *Journal of Assisted Reproduction and Genetics*, **2009**, 26, 113-7	3.4	5
40	Anomalies gh†tiques et infertilit˜masculine. *Andrologie*, **2009**, 19, 2-16		2
39	Fetal karyotype in feto-fetal transfusion syndrome: a 7-year experience. *Prenatal Diagnosis*, **2009**, 29, 804-5	3.2	1
38	Sperm chromosome analysis of an infertile patient with a 95% mosaic r(21) karyotype and normal phenotype. *Fertility and Sterility*, **2009**, 91, 930.e13-5	4.8	10
37	Is classic pericentric inversion of chromosome 2 inv(2)(p11q13) associated with an increased risk of unbalanced chromosomes?. *Fertility and Sterility*, **2009**, 92, 1497.e1-1497.e4	4.8	8
36	Meiotic segregation of X-autosome translocation in two carriers and implications for assisted reproduction. *Reproductive BioMedicine Online*, **2009**, 18, 850-5	4	5
35	Pronuclear morphology differs between women more than 38 and women less than 30 years of age. *Reproductive BioMedicine Online*, **2009**, 18, 367-73	4	8
34	Case report: Meiotic segregation in spermatozoa of a 46,X,t(Y;10)(q11.2;p15.2) fertile translocation carrier. *Reproductive BioMedicine Online*, **2009**, 18, 549-54	4	6
33	Array comparative genomic hybridization in prenatal diagnosis: another experience. *Fetal Diagnosis and Therapy*, **2009**, 25, 277-84	2.4	63

32	Partial chromosome deletion: a new trisomy rescue mechanism?. *Fetal Diagnosis and Therapy*, **2009**, 25, 111-4	2.4	4
31	Stability of aneuploidy rate in polar bodies in two cohorts from the same patient. *Reproductive BioMedicine Online*, **2008**, 17, 213-9	4	5
30	Tumor necrosis factor-alpha -308 polymorphism in infertile men with altered sperm production or motility. *Human Reproduction*, **2008**, 23, 2858-66	5.7	26
29	How viable are zygotes in which the PN are still intact at 25 hours? Impact on the choice of embryo for transfer. *Fertility and Sterility*, **2008**, 90, 551-6	4.8	5
28	Status of the executioner step of apoptosis in human with normal spermatogenesis and azoospermia. *Fertility and Sterility*, **2008**, 90, 1723-31	4.8	17
27	Gamete cytogenetic study in couples with implantation failure: aneuploidy rate is increased in both couple members. *Journal of Assisted Reproduction and Genetics*, **2008**, 25, 539-45	3.4	19
26	Observation des spermatozoïdes au fort grossissement (MSOME): intérêt et perspectives. *Andrologie*, **2008**, 18, 26-34		
25	La sélection des spermatozoïdes à fort grossissement permet-elle une diminution de la fréquence des aneuploïdies spermatiques?. *Andrologie*, **2008**, 18, 274-287		2
24	Evaluation de l'apport de la méthode d'observation des spermatozoïdes à fort grossissement en ICSI. *Andrologie*, **2007**, 17, 212-222		11
23	Sperm chromosome analysis in two cases of paracentric inversion. *Fertility and Sterility*, **2007**, 87, 418.e1-4	4.8	9
22	Oocyte aneuploidy mechanisms are different in two situations of increased chromosomal risk: older patients and patients with recurrent implantation failure after in vitro fertilization. *Fertility and Sterility*, **2007**, 87, 1333-9	4.8	24
21	Evidence of a high proportion of premature unbalanced separation of sister chromatids in the first polar bodies of women of advanced age. *Human Reproduction*, **2006**, 21, 1172-8	5.7	47
20	Chromosomal analysis of spermatozoa with normal-sized heads in two infertile patients with macrocephalic sperm head syndrome. *Fertility and Sterility*, **2006**, 85, 750.e5-750.e7	4.8	28
19	Whole-arm translocations between chromosome 1 and acrocentric G chromosomes are associated with a poor prognosis for spermatogenesis: two new cases and review of the literature. *Fertility and Sterility*, **2006**, 86, 1001.e1-5	4.8	6
18	Partial trisomy 20p resulting from recombination of a maternal pericentric inversion: case report of a prenatal diagnosis by chorionic villus sampling. *Prenatal Diagnosis*, **2006**, 26, 239-41	3.2	9
17	Karyotype and outcome of fetuses diagnosed with cystic hygroma in the first trimester in relation to nuchal translucency thickness. *Prenatal Diagnosis*, **2006**, 26, 369-72	3.2	34
16	Screening for Down syndrome using first-trimester combined screening followed by second-trimester ultrasound examination in an unselected population. *American Journal of Obstetrics and Gynecology*, **2006**, 195, 1379-87	6.4	66
15	A histomorphometric and cytogenetic study of testis from men 29-102 years old. *Fertility and Sterility*, **2005**, 83, 923-8	4.8	42

14	Le risque chromosomique pour un patient porteur d'une translocation t(X;2) concerne non seulement la translocation mais aussi la ségrégation XY. *Andrologie*, **2005**, 15, 328-333		3
13	Prenatal diagnosis of de novo (7;19)(q11.2;q13.3) translocation associated with a thick corpus callosum and Wilms tumor of the kidneys. *Prenatal Diagnosis*, **2005**, 25, 876-8	3.2	10
12	Dandy-Walker syndrome and corpus callosum agenesis in 5p deletion. *Prenatal Diagnosis*, **2005**, 25, 311-3	3.2	23
11	Functional disomy of Xp including duplication of DAX1 gene with sex reversal due to t(X;Y)(p21.2;p11.3) **2004**, 128A, 325-30		23
10	Mechanism of intrachromosomal triplications 15q11-q13: a new clinical report. *American Journal of Medical Genetics Part A*, **2003**, 118A, 229-34		20
9	Possible human chimera detected prenatally after in vitro fertilization: a case report. *Prenatal Diagnosis*, **2003**, 23, 935-7	3.2	16
8	Expression of the cystathionine beta synthase (CBS) gene during mouse development and immunolocalization in adult brain. *Journal of Histochemistry and Cytochemistry*, **2003**, 51, 363-71	3.4	117
7	Fetal gender: antenatal discrepancy between phenotype and genotype. *Ultrasound in Obstetrics and Gynecology*, **2002**, 20, 286-9	5.8	10
6	The X chromosome and ovarian function. *Cytogenetic and Genome Research*, **2002**, 99, 218-23	1.9	5
5	Deletion of 9p associated with gonadal dysfunction in 46,XY but not in 46,XX human fetuses. *Journal of Medical Genetics*, **2002**, 39, 514-8	5.8	27
4	Pierre Robin sequence and interstitial deletion 2q32.3-q33.2. *American Journal of Medical Genetics Part A*, **2001**, 102, 219-26		23
3	Molecular cytogenetic analysis of a duplication Xp in a female with an abnormal phenotype and random X inactivation. *Clinical Genetics*, **2000**, 58, 116-22	4	19
2	Overexpression of mSim2 gene in the zona limitans of the diencephalon of segmental trisomy 16 Ts1Cje fetuses, a mouse model for trisomy 21: a novel whole-mount based RNA hybridization study. *Developmental Brain Research*, **2000**, 121, 73-8		13
1	Characterization of a novel gene, C21orf6, mapping to a critical region of chromosome 21q22.1 involved in the monosomy 21 phenotype and of its murine ortholog, orf5. *Genomics*, **2000**, 64, 203-10	4.3	3