Sharon E Plon

List of Publications by Year
in descending order

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128 papers	12,646 citations	38720 50 h-index	27389 106 g-index
138 all docs	138 docs citations	138 times ranked	21209 citing authors

#	Article	IF	Citations
1	Distinct somatic DICER1 hotspot mutations in three metachronous ovarian Sertoli-Leydig cell tumors in a patient with DICER1 syndrome. Cancer Genetics, 2022, 262-263, 53-56.	0.2	2
2	Standards for the classification of pathogenicity of somatic variants in cancer (oncogenicity): Joint recommendations of Clinical Genome Resource (ClinGen), Cancer Genomics Consortium (CGC), and Variant Interpretation for Cancer Consortium (VICC). Genetics in Medicine, 2022, 24, 986-998.	1.1	55
3	ClinGen Variant Curation Interface: a variant classification platform for the application of evidence criteria from ACMG/AMP guidelines. Genome Medicine, 2022, 14, 6.	3.6	34
4	The annual ASHG dinner. American Journal of Human Genetics, 2022, 109, 377-378.	2.6	1
5	A Validation Framework for Somatic Copy Number Detection in Targeted Sequencing Panels. Journal of Molecular Diagnostics, 2022, 24, 760-774.	1.2	3
6	LGG-04. Clinical and molecular characterization of metastatic pediatric low grade gliomas. Neuro-Oncology, 2022, 24, i87-i87.	0.6	0
7	Clinical and molecular features of pediatric cancer patients with Lynch syndrome. Pediatric Blood and Cancer, 2022, 69, .	0.8	6
8	Clinical and functional characterization of telomerase variants in patients with pediatric acute myeloid leukemia/myelodysplastic syndrome. Leukemia, 2021, 35, 269-273.	3.3	4
9	Specifications of the ACMG/AMP variant interpretation guidelines for germline TP53 variants. Human Mutation, 2021, 42, 223-236.	1.1	81
10	Pediatric Rhabdomyosarcoma: Epidemiology and Genetic Susceptibility. Journal of Clinical Medicine, 2021, 10, 2028.	1.0	25
11	Genomic analysis and preclinical xenograft model development identify potential therapeutic targets for MYOD1 â€mutant softâ€tissue sarcoma of childhood. Journal of Pathology, 2021, 255, 52-61.	2.1	3
12	Pediatric Oncologistsâ€™ Experiences Returning and Incorporating Genomic Sequencing Results into Cancer Care. Journal of Personalized Medicine, 2021, 11, 570.	1.1	2
13	Durable Response to Larotrectinib in a Child With Histologic Diagnosis of Recurrent Disseminated Ependymoma Discovered to Harbor an NTRK2 Fusion: The Impact of Integrated Genomic Profiling. JCO Precision Oncology, 2021, 5, 1221-1227.	1.5	5
14	Germline Cancer Predisposition Variants inâ€, Pediatric Rhabdomyosarcoma: A Report From the Childrenâ€™s Oncology Group. Journal of the National Cancer Institute, 2021, 113, 875-883.	3.0	55
15	Importance of Population-Based Cancer Risk Information in the Care of Patients With Rare Genetic Disorders. Journal of Clinical Oncology, 2021, , JCO2102251.	0.8	0
16	A brief history of human disease genetics. Nature, 2020, 577, 179-189.	13.7	441
17	Male Sex and the Risk of Childhood Cancer: The Mediating Effect of Birth Defects. JNCI Cancer Spectrum, 2020, 4, pkaa052.	1.4	7
18	Points to consider for reporting of germline variation in patients undergoing tumor testing: a statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2020, 22, 1142-1148.	1.1	59

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19	Ancestry-specific predisposing germline variants in cancer. Genome Medicine, 2020, 12, 51.	3.6	35
20	Cancer diagnostic profile in children with structural birth defects: An assessment in 15,000 childhood cancer cases. Cancer, 2020, 126, 3483-3492.	2.0	12
21	Clinical Genetics Lacks Standard Definitions and Protocols for the Collection and Use of Diversity Measures. American Journal of Human Genetics, 2020, 107, 72-82.	2.6	52
22	EPEN-25. EXCEPTIONAL CLINICAL AND IMAGING RESPONSE TO TRK-INHIBITION IN A PATIENT WITH SUPRATENTORIAL EPENDYMOMA HARBORING NTRK2 GENE FUSION. Neuro-Oncology, 2020, 22, iii312-iii313.	0.6	0
23	PATH-27. MUTATION DETECTION USING PLASMA CELL-FREE DNA IN CHILDREN WITH CENTRAL NERVOUS SYSTEM TUMORS. Neuro-Oncology, 2020, 22, iii430-iii430.	0.6	0
24	PATH-29. HIGH FREQUENCY OF CLINICALLY-RELEVANT TUMOR VARIANTS DETECTED BY MOLECULAR TESTING OF HIGH-RISK PEDIATRIC CNS TUMORS â€“ PRELIMINARY FINDINGS FROM THE TEXAS KidsCanSeq STUDY. Neuro-Oncology, 2020, 22, iii430-iii430.	0.6	0
25	Genome-wide sequencing in acutely ill infants: genomic medicineâ€™s critical application?. Genetics in Medicine, 2019, 21, 498-504.	1.1	42
26	Mutations in ANAPC1, Encoding a Scaffold Subunit of the Anaphase-Promoting Complex, Cause Rothmund-Thomson Syndrome Type 1. American Journal of Human Genetics, 2019, 105, 625-630.	2.6	42
27	Molecular profiling predicts meningioma recurrence and reveals loss of DREAM complex repression in aggressive tumors. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 21715-21726.	3.3	122
28	Association Between Birth Defects and Cancer Risk Among Children and Adolescents in a Population-Based Assessment of 10 Million Live Births. JAMA Oncology, 2019, 5, 1150.	3.4	87
29	Responsibility, culpability, and parental views on genomic testing for seriously ill children. Genetics in Medicine, 2019, 21, 2791-2797.	1.1	20
30	GENE-09. MUTATION SIGNATURE ANALYSIS IN AN ULTRAHYPERMUTATED MEDULLOBLASTOMA PREDICTS UNDERLYING GERMLINE POLYMERASE PROOFREADING DEFICIENCY IN A CHILD WITH CLINICAL FEATURES OF CONSTITUTIONAL MISMATCH REPAIR DEFICIENCY SYNDROME. Neuro-Oncology, 2019, 21, ii82-ii83.	0.6	0
31	Genetic Predisposition to Childhood Cancer in the Genomic Era. Annual Review of Genomics and Human Genetics, 2019, 20, 241-263.	2.5	27
32	Severe therapyâ€related toxicities after treatment for Hodgkin lymphoma due to a pathogenic TERT variant and shortened telomeres. Pediatric Blood and Cancer, 2019, 66, e27779.	0.8	3
33	Characterization of pediatric hepatocellular carcinoma reveals genomic heterogeneity and diverse signaling pathway activation. Pediatric Blood and Cancer, 2019, 66, e27745.	0.8	37
34	Germline POLE mutation in a child with hypermutated medulloblastoma and features of constitutional mismatch repair deficiency. Journal of Physical Education and Sports Management, 2019, 5, a004499.	0.5	19
35	ClinGen Myeloid Malignancy Variant Curation Expert Panel recommendations for germline RUNX1 variants. Blood Advances, 2019, 3, 2962-2979.	2.5	110
36	A case for expert curation: an overview of cancer curation in the Clinical Genome Resource (ClinGen). Journal of Physical Education and Sports Management, 2019, 5, a004739.	0.5	14

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37	Agents of empathy: How medical interpreters bridge sociocultural gaps in genomic sequencing disclosures with Spanish-speaking families. Patient Education and Counseling, 2019, 102, 895-901.	1.0	14
38	Exome sequencing disclosures in pediatric cancer care: Patterns of communication among oncologists, genetic counselors, and parents. Patient Education and Counseling, 2019, 102, 680-686.	1.0	12
39	Current Controversies in Prenatal Diagnosis 2: NIPT results suggesting maternal cancer should always be disclosed. Prenatal Diagnosis, 2019, 39, 339-343.	1.1	31
40	Determining the clinical validity of hereditary colorectal cancer and polyposis susceptibility genes using the Clinical Genome Resource Clinical Validity Framework. Genetics in Medicine, 2019, 21, 1507-1516.	1.1	19
41	Clinical validity assessment of genes frequently tested on hereditary breast and ovarian cancer susceptibility sequencing panels. Genetics in Medicine, 2019, 21, 1497-1506.	1.1	52
42	Framework for microRNA variant annotation and prioritization using human population and disease datasets. Human Mutation, 2019, 40, 73-89.	1.1	18
43	Secondary findings from clinical genomic sequencing: prevalence, patient perspectives, family history assessment, and health-care costs from a multisite study. Genetics in Medicine, 2019, 21, 1100-1110.	1.1	111
44	Pathogenic Germline Variants in 10,389 Adult Cancers. Cell, 2018, 173, 355-370.e14.	13.5	620
45	Key Implications of Data Sharing in Pediatric Genomics. JAMA Pediatrics, 2018, 172, 476.	3.3	29
46	ClinGen Allele Registry links information about genetic variants. Human Mutation, 2018, 39, 1690-1701.	1.1	48
47	BRCA Challenge: BRCA Exchange as a global resource for variants in BRCA1 and BRCA2. PLoS Genetics, 2018, 14, e1007752.	1.5	148
48	The clinical imperative for inclusivity: Race, ethnicity, and ancestry (REA) in genomics. Human Mutation, 2018, 39, 1713-1720.	1.1	102
49	Geneâ€specific criteria for PTEN variant curation: Recommendations from the ClinGen PTEN Expert Panel. Human Mutation, 2018, 39, 1581-1592.	1.1	123
50	Updated recommendation for the benign standâ€alone ACMG/AMP criterion. Human Mutation, 2018, 39, 1525-1530.	1.1	102
51	Specifications of the ACMG/AMP variant curation guidelines for the analysis of germline CDH1 sequence variants. Human Mutation, 2018, 39, 1553-1568.	1.1	138
52	Integrating somatic variant data and biomarkers for germline variant classification in cancer predisposition genes. Human Mutation, 2018, 39, 1542-1552.	1.1	40
53	ClinGen Variant Curation Expert Panel experiences and standardized processes for disease and geneâ€level specification of the ACMG/AMP guidelines for sequence variant interpretation. Human Mutation, 2018, 39, 1614-1622.	1.1	132
54	ClinGen and ClinVar â€“ Enabling Genomics in Precision Medicine. Human Mutation, 2018, 39, 1473-1475.	1.1	14

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55	The Clinical Sequencing Evidence-Generating Research Consortium: Integrating Genomic Sequencing in Diverse and Medically Underserved Populations. American Journal of Human Genetics, 2018, 103, 319-327.	2.6	122
56	TBIO-20. CLINICAL TUMOR WHOLE EXOME SEQUENCING FOR PEDIATRIC NEURO-ONCOLOGY PATIENTS â€“ RESULTS FROM THE BAYLOR ADVANCING SEQUENCING IN CHILDHOOD CANCER CARE (BASIC3) CLINICAL SEQUENCING STUDY. Neuro-Oncology, 2018, 20, i184-i184.	0.6	0
57	The Ancestral Pace of Variant Reclassification. Journal of the National Cancer Institute, 2018, 110, 1133-1134.	3.0	7
58	Abstract 5359: Regulatory germline variants in 10,389 adult cancers. Cancer Research, 2018, 78, 5359-5359.	0.4	13
59	Myeloid Malignancy Variant Curation Expert Panel: An ASH-Sponsored Clingen Expert Panel to Optimize and Validate Acmg/AMP Variant Interpretation Guidelines for Genes Associated with Inherited Myeloid Neoplasms. Blood, 2018, 132, 5849-5849.	0.6	0
60	ClinGen Pathogenicity Calculator: a configurable system for assessing pathogenicity of genetic variants. Genome Medicine, 2017, 9, 3.	3.6	59
61	Cancer and Central Nervous System Tumor Surveillance in Pediatric Neurofibromatosis 1. Clinical Cancer Research, 2017, 23, e46-e53.	3.2	133
62	Recommendations for Childhood Cancer Screening and Surveillance in DNA Repair Disorders. Clinical Cancer Research, 2017, 23, e23-e31.	3.2	93
63	Clinical Management and Tumor Surveillance Recommendations of Inherited Mismatch Repair Deficiency in Childhood. Clinical Cancer Research, 2017, 23, e32-e37.	3.2	157
64	Pediatric Cancer Predisposition and Surveillance: An Overview, and a Tribute to Alfred G. Knudson Jr. Clinical Cancer Research, 2017, 23, e1-e5.	3.2	130
65	Evaluating the Clinical Validity of Gene-Disease Associations: An Evidence-Based Framework Developed by the Clinical Genome Resource. American Journal of Human Genetics, 2017, 100, 895-906.	2.6	403
66	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	3.6	184
67	Germline mutations in ABL1 cause an autosomal dominant syndrome characterized by congenital heart defects and skeletal malformations. Nature Genetics, 2017, 49, 613-617.	9.4	40
68	Resolution of Disease Phenotypes Resulting from Multilocus Genomic Variation. New England Journal of Medicine, 2017, 376, 21-31.	13.9	565
69	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	3.3	348
70	Haploinsufficiency of the Chromatin Remodeler BPTF Causes Syndromic Developmental and Speech Delay, Postnatal Microcephaly, and Dysmorphic Features. American Journal of Human Genetics, 2017, 101, 503-515.	2.6	61
71	Surveillance Recommendations for Children with Overgrowth Syndromes and Predisposition to Wilms Tumors and Hepatoblastoma. Clinical Cancer Research, 2017, 23, e115-e122.	3.2	140
72	Cancer Screening Recommendations and Clinical Management of Inherited Gastrointestinal Cancer Syndromes in Childhood. Clinical Cancer Research, 2017, 23, e107-e114.	3.2	91

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73	A survey of current practices for genomic sequencing test interpretation and reporting processes in US laboratories. Genetics in Medicine, 2017, 19, 575-582.	1.1	68
74	Renal cell carcinoma harboring somatic TSC2 mutations in a child with methylmalonic acidemia. Pediatric Blood and Cancer, 2017, 64, e26286.	0.8	9
75	Portero versus portador: Spanish interpretation of genomic terminology during whole exome sequencing results disclosure. Personalized Medicine, 2017, 14, 503-514.	0.8	17
76	Parental Perspectives on Whole-Exome Sequencing in Pediatric Cancer: A Typology of Perceived Utility. JCO Precision Oncology, 2017, 1, 1-10.	1.5	26
77	Evaluation of in silico algorithms for use with ACMG/AMP clinical variant interpretation guidelines. Genome Biology, 2017, 18, 225.	3.8	185
78	Childhood cancer risk in those with chromosomal and non-chromosomal congenital anomalies in Washington State: 1984-2013. PLoS ONE, 2017, 12, e0179006.	1.1	36
79	Family-based exome-wide association study of childhood acute lymphoblastic leukemia among Hispanics confirms role of ARID5B in susceptibility. PLoS ONE, 2017, 12, e0180488.	1.1	13
80	Current state of pediatric sarcoma biology and opportunities for future discovery: A report from the sarcoma translational research workshop. Cancer Genetics, 2016, 209, 182-194.	0.2	38
81	Performance of ACMG-AMP Variant-Interpretation Guidelines among Nine Laboratories in the Clinical Sequencing Exploratory Research Consortium. American Journal of Human Genetics, 2016, 98, 1067-1076.	2.6	432
82	Recommendations for the integration of genomics into clinical practice. Genetics in Medicine, 2016, 18, 1075-1084.	1.1	125
83	Integrated tumor and germline whole-exome sequencing identifies mutations in MAPK and PI3K pathway genes in an adolescent with rosette-forming glioneuronal tumor of the fourth ventricle. Journal of Physical Education and Sports Management, 2016, 2, a001057.	0.5	21
84	Familyâ€based exomeâ€wide assessment of maternal genetic effects on susceptibility to childhood Bâ€cell acute lymphoblastic leukemia in hispanics. Cancer, 2016, 122, 3697-3704.	2.0	15
85	Improvement of outcomes for TP53 carriers. Lancet Oncology, The, 2016, 17, 1184-1186.	5.1	7
86	Mutations in the nuclear bile acid receptor FXR cause progressive familial intrahepatic cholestasis. Nature Communications, 2016, 7, 10713.	5.8	227
87	SV-STAT accurately detects structural variation via alignment to reference-based assemblies. Source Code for Biology and Medicine, 2016, 11, 8.	1.7	3
88	Is Whole-Exome Sequencing an Ethically Disruptive Technology? Perspectives of Pediatric Oncologists and Parents of Pediatric Patients With Solid Tumors. Pediatric Blood and Cancer, 2016, 63, 511-515.	0.8	39
89	ITD assembler: an algorithm for internal tandem duplication discovery from short-read sequencing data. BMC Bioinformatics, 2016, 17, 188.	1.2	16
90	Shorter Remission Telomere Length Predicts Delayed Neutrophil Recovery After Acute Myeloid Leukemia Therapy: A Report From the Childrenâ€™s Oncology Group. Journal of Clinical Oncology, 2016, 34, 3766-3772.	0.8	17

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91	Diagnostic Yield of Clinical Tumor and Germline Whole-Exome Sequencing for Children With Solid Tumors. JAMA Oncology, 2016, 2, 616.	3.4	378
92	Molecular diagnostic experience of whole-exome sequencing in adult patients. Genetics in Medicine, 2016, 18, 678-685.	1.1	186
93	Germline Findings in Tumor-Only Sequencing: Points to Consider for Clinicians and Laboratories: Table 1.. Journal of the National Cancer Institute, 2016, 108, djv351.	3.0	86
94	PMS2 monoallelic mutation carriers: the known unknown. Genetics in Medicine, 2016, 18, 13-19.	1.1	51
95	Hepatocellular carcinoma associated with tightâ€junction protein 2 deficiency. Hepatology, 2015, 62, 1914-1916.	3.6	63
96	Family history of cancer and childhood rhabdomyosarcoma: a report from the Children's Oncology Group and the Utah Population Database. Cancer Medicine, 2015, 4, 781-790.	1.3	25
97	ClinGen â€" The Clinical Genome Resource. New England Journal of Medicine, 2015, 372, 2235-2242.	13.9	1,016
98	Germline Mutations in Shelterin Complex Genes Are Associated With Familial Glioma. Journal of the National Cancer Institute, 2015, 107, 384.	3.0	172
99	Identifying gene disruptions in novel balanced de novo constitutional translocations in childhood cancer patients by whole-genome sequencing. Genetics in Medicine, 2015, 17, 831-835.	1.1	7
100	Targeted Sequencing in Chromosome 17q Linkage Region Identifies Familial Glioma Candidates in the Gliogene Consortium. Scientific Reports, 2015, 5, 8278.	1.6	22
101	CSER and eMERGE: current and potential state of the display of genetic information in the electronic health record. Journal of the American Medical Informatics Association: JAMIA, 2015, 22, 1231-1242.	2.2	73
102	A Comprehensive Strategy for Accurate Mutation Detection of the Highly Homologous PMS2. Journal of Molecular Diagnostics, 2015, 17, 545-553.	1.2	42
103	BCORâ€“CCNB3 fusions are frequent in undifferentiated sarcomas of male children. Modern Pathology, 2015, 28, 575-586.	2.9	122
104	Obtaining informed consent for clinical tumor and germline exome sequencing of newly diagnosed childhood cancer patients. Genome Medicine, 2014, 6, 69.	3.6	60
105	Molecular Findings Among Patients Referred for Clinical Whole-Exome Sequencing. JAMA - Journal of the American Medical Association, 2014, 312, 1870.	3.8	1,171
106	De Novo Truncating Mutations in AHDC1 in Individuals with Syndromic Expressive Language Delay, Hypotonia, and Sleep Apnea. American Journal of Human Genetics, 2014, 94, 784-789.	2.6	57
107	A recurrent germline PAX5 mutation confers susceptibility to pre-B cell acute lymphoblastic leukemia. Nature Genetics, 2013, 45, 1226-1231.	9.4	270
108	Identification of TP53 as an acute lymphocytic leukemia susceptibility gene through exome sequencing. Pediatric Blood and Cancer, 2013, 60, E1-3.	0.8	44

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109	Processes and preliminary outputs for identification of actionable genes as incidental findings in genomic sequence data in the Clinical Sequencing Exploratory Research Consortium. Genetics in Medicine, 2013, 15, 860-867.	1.1	99
110	Implementation and evaluation of clinical exome sequencing in childhood cancer care: The BASIC3 study.. Journal of Clinical Oncology, 2013, 31, 10023-10023.	0.8	1
111	Constitutional tandem duplication of 9q34 that truncates EHMT1 in a child with ganglioglioma. Pediatric Blood and Cancer, 2012, 58, 801-805.	0.8	8
112	Identification of genetic susceptibility to childhood cancer through analysis of genes in parallel. Cancer Genetics, 2011, 204, 19-25.	0.2	14
113	Prediction of missense mutation functionality depends on both the algorithm and sequence alignment employed. Human Mutation, 2011, 32, 661-668.	1.1	195
114	Genetic testing and cancer risk management recommendations by physicians for at-risk relatives. Genetics in Medicine, 2011, 13, 148-154.	1.1	99
115	Unifying cancer genetics. Genetics in Medicine, 2011, 13, 203-204.	1.1	1
116	Clinical and Molecular Characterization of Germline Telomerase Reverse Transcriptase (TERT) Variants in Children with Acute Myeloid Leukemia (AML),. Blood, 2011, 118, 3571-3571.	0.6	0
117	Overexpression of ZNF342 by juxtaposition with MPO promoter/enhancer in the novel translocation t(17;19)(q23;q13.32) in pediatric acute myeloid leukemia and analysis of ZNF342 expression in leukemia. Genes Chromosomes and Cancer, 2009, 48, 480-489.	1.5	14
118	Design and analysis of epidemiological studies of excess cancer among children exposed to chernobyl radionuclides. Stem Cells, 2009, 15, 211-230.	1.4	4
119	Sequence variant classification and reporting: recommendations for improving the interpretation of cancer susceptibility genetic test results. Human Mutation, 2008, 29, 1282-1291.	1.1	782
120	Heterozygous Screen in Saccharomyces cerevisiae Identifies Dosage-Sensitive Genes That Affect Chromosome Stability. Genetics, 2008, 178, 1193-1207.	1.2	24
121	Prevalence and Clinical Correlates of JAK2 Mutations in Pediatric Down Syndrome Acute Lymphoblastic Leukemia.. Blood, 2008, 112, 1506-1506.	0.6	1
122	Nuclear import and retention domains in the amino terminus of RECQL4. Gene, 2007, 391, 26-38.	1.0	44
123	A constitutional balanced t(3;8)(p14;q24.1) translocation results in disruption of theTRC8 gene and predisposition to clear cell renal cell carcinoma. Genes Chromosomes and Cancer, 2007, 46, 805-812.	1.5	31
124	Clinical Utility of Array Comparative Genomic Hybridization for Detection of Chromosomal Abnormalities in Pediatric Acute Lymphoblastic Leukemia.. Blood, 2006, 108, 2275-2275.	0.6	9
125	Inherited Susceptibility for Pediatric Cancer. Cancer Journal (Sudbury, Mass), 2005, 11, 255-267.	1.0	23
126	FBN1 exon 2 splicing error in a patient with Marfan syndrome. American Journal of Medical Genetics Part A, 2001, 101, 130-134.	2.4	16

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127	Clinical manifestations in a cohort of 41 Rothmund-Thomson syndrome patients. American Journal of Medical Genetics Part A, 2001, 102, 11-17.	2.4	290
128	Neoplasms in neurofibromatosis 1 are related to gender but not to family history of cancer. Genetic Epidemiology, 2001, 20, 75-86.	0.6	24