

## Svetlana A Yatsenko

Professor of Pathology

### Bio

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#### ACADEMIC APPOINTMENTS

- Professor - University Medical Line, Pathology
- Member, Maternal & Child Health Research Institute (MCHRI)

### Publications

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#### PUBLICATIONS

- **Unusual Recombinant Chromosome 6 Derived From a Parental Rearrangement With Complex Paracentric Inversions.** *American journal of medical genetics. Part A*  
Babcock, M., Daghani, M., Sebastian, J., Lancaster, E., Ghaloul-Gonzalez, L., Ortiz, D., Powell, E., Bellissimo, D. B., Sahoo, T., Yatsenko, S. A.  
2026
- **The Human Intolerome: a curated database to prioritize genomic variants in stillbirth, pregnancy loss, and neonatal death.** *Genetics in medicine : official journal of the American College of Medical Genetics*  
Yatsenko, S. A., Nagasuri, A., Soman, V., Garakani, A., Aminbeidokhti, M., Chandran, U., Aarabi, M., Oskotsky, T., Bukhari, S. H., Hypes, C., Gu, Z., Monteiro, R., Smith, et al  
2026: 102546
- **Genetics of Primary Ovarian Insufficiency.** *Seminars in reproductive medicine*  
Yatsenko, S. A., Rajkovic, A.  
2026
- **Discrepancies Between Sex Prediction and Fetal Sex After Prenatal Noninvasive Cell-Free DNA Screening.** *Journal of the Endocrine Society*  
Witchel, S. F., Rajkovic, A., Yatsenko, S. A.  
2025; 9 (2): bvaf007
- **Miscarriage risk assessment: a bioinformatic approach to identifying candidate lethal genes and variants.** *Human genetics*  
Aminbeidokhti, M., Qu, J., Belur, S., Cakmak, H., Jaswa, E., Lathi, R. B., Sirota, M., Snyder, M. P., Yatsenko, S. A., Rajkovic, A.  
2024
- **A Novel Integrated Approach for Cytogenomic Evaluation of Plasma Cell Neoplasms.** *The Journal of molecular diagnostics : JMD*  
Aarabi, M., Yoest, J. M., Farah, R., Rajkovic, A., Swerdlow, S. H., Yatsenko, S. A.  
2022; 24 (10): 1067-1078
- **Carrier frequency of autosomal recessive genetic conditions in diverse populations: Lessons learned from the genome aggregation database.** *Clinical genetics*  
Schmitz, M. J., Aarabi, M., Bashar, A., Rajkovic, A., Gregg, A. R., Yatsenko, S. A.  
2022; 102 (2): 87-97
- **Reproductive outcomes in individuals with chromosomal reciprocal translocations.** *Genetics in medicine : official journal of the American College of Medical Genetics*  
Verdoni, A., Hu, J., Surti, U., Babcock, M., Sheehan, E., Clemens, M., Drewes, S., Walsh, L., Clark, R., Katari, S., Sanfilippo, J., Saller, D. N., Rajkovic, et al  
2021; 23 (9): 1753-1760
- **Cytogenetic signatures of recurrent pregnancy losses.** *Prenatal diagnosis*

- Yatsenko, S. A., Quesada-Candela, C., Saller, D. N., Beck, S., Jaffe, R., Kostadinov, S., Yanowitz, J., Rajkovic, A.  
2021; 41 (1): 70-78
- **Testis formation in XX individuals resulting from novel pathogenic variants in Wilms' tumor 1 (WT1) gene.** *Proceedings of the National Academy of Sciences of the United States of America*  
Eozenou, C., Gonen, N., Touzon, M. S., Jorgensen, A., Yatsenko, S. A., Fusee, L., Kamel, A. K., Gellen, B., Guercio, G., Singh, P., Witchel, S., Berman, A. J., Mainpal, et al  
2020; 117 (24): 13680-13688
  - **A high-resolution X chromosome copy-number variation map in fertile females and women with primary ovarian insufficiency.** *Genetics in medicine : official journal of the American College of Medical Genetics*  
Yatsenko, S. A., Wood-Trageser, M., Chu, T., Jiang, H., Rajkovic, A.  
2019; 21 (10): 2275-2284
  - **Female-to-male sex reversal associated with unique Xp21.2 deletion disrupting genomic regulatory architecture of the dosage-sensitive sex reversal region.** *Journal of medical genetics*  
Dangle, P., Touzon, M. S., Reyes-Múgica, M., Witchel, S. F., Rajkovic, A., Schneck, F. X., Yatsenko, S. A.  
2017; 54 (10): 705-709
  - **Maternal cell-free DNA-based screening for fetal microdeletion and the importance of careful diagnostic follow-up.** *Genetics in medicine : official journal of the American College of Medical Genetics*  
Yatsenko, S. A., Peters, D. G., Saller, D. N., Chu, T., Clemens, M., Rajkovic, A.  
2015; 17 (10): 836-8
  - **X-linked TEX11 mutations, meiotic arrest, and azoospermia in infertile men.** *The New England journal of medicine*  
Yatsenko, A. N., Georgiadis, A. P., Röpke, A., Berman, A. J., Jaffe, T., Olszewska, M., Westernströer, B., Sanfilippo, J., Kurpisz, M., Rajkovic, A., Yatsenko, S. A., Kliesch, S., Schlatt, et al  
2015; 372 (22): 2097-107
  - **Assessing patient perceptions and understandings of genetic testing after pregnancy loss.** *Journal of assisted reproduction and genetics*  
Hrach, E., Carlson, J., Grubs, R. E., Sheehan, E., Lathi, R. B., Rajkovic, A., Yatsenko, S. A.  
2025
  - **Molecular taxonomy of MDS/CMML patients influences responses to hypomethylating agents and clinical outcomes.** *Leukemia research*  
Molina, A., Khanna, V., Jensen, A., Stehr, H., Tan, B., Yatsenko, S., Greenberg, P. L.  
2025; 156: 107736
  - **Myeloid neoplasms with oligomonocytosis exhibit heterogenous pathologic and genetic features.** *Modern pathology : an official journal of the United States and Canadian Academy of Pathology, Inc*  
Baloda, V., Al Amri, R., Patwardhan, P. P., Monaghan, S. A., Moore, E. M., Rea, B., Djokic, M., Aggarwal, N., Bullock, G. C., Liu, Y., Yatsenko, S., Bailey, N. G.  
2025: 100823
  - **Recurrent Xp22.31-Yq11 Unbalanced Translocations: Molecular Diagnosis and Clinical Implications in Three Families.** *American journal of medical genetics. Part A*  
Daghsni, M., Sheehan, E., Madan-Khetarpal, S., Aarabi, M., Witchel, S. F., Rajkovic, A., Yatsenko, S. A.  
2024: e63913
  - **Hiding in Plain Sight: Radiologic and Pathologic Findings Can Identify Beckwith-Wiedemann Syndrome in Patients With Wilms Tumor.** *Journal of pediatric hematology/oncology*  
Molina, L. M., Rao, A., Meade, J., Squires, J. H., Yatsenko, S. A., Salgado, C. M., Reyes-Mugica, M.  
2024
  - **ASSESSING PATIENT PERCEPTIONS AND UNDERSTANDINGS OF GENETIC TESTING AFTER PREGNANCY LOSS**  
Hrach, E., Carlson, J., Grubs, R. E., Sheehan, E., Lathi, R. B., Rajkovic, A., Yatsenko, S.  
ELSEVIER SCIENCE INC.2024: E111
  - **Decoding the Mystery of Recurrent Pregnancy Loss: Revelations from Genome Sequencing.**  
Aminbeidokhti, M., Nagasuri, A., Cakmak, H., Jaswa, E., Baldwin, M., Edelman, A., Pollard, E., Snyder, M., Sirota, M., Tise, C., Bernstein, J., Stephenson, M., Lathi, et al  
SPRINGER HEIDELBERG.2024: 76A

- **Single-cell morphological and transcriptome analysis unveil inhibitors of polyploid giant breast cancer cells in vitro.** *Communications biology*  
Zhou, M., Ma, Y., Chiang, C. C., Rock, E. C., Butler, S. C., Anne, R., Yatsenko, S., Gong, Y., Chen, Y. C.  
2023; 6 (1): 1301
- **Novel FIP1L1::KIT fusion in a myeloid neoplasm with eosinophilia, T-lymphoblastic transformation, and dasatinib response.** *Haematologica*  
Alsouqi, A., Kleinberger, J., Werner, T. S., Awan, R., Chopra, S., Rea, B., Aggarwal, N., Yatsenko, S. A., Farah, R., Bailey, N. G.  
2023; 108 (11): 3181-3185
- **A Multicenter Analysis of Abnormal Chromosomal Microarray Findings in Congenital Heart Disease.** *Journal of the American Heart Association*  
Landis, B. J., Helvaty, L. R., Geddes, G. C., Lin, J. I., Yatsenko, S. A., Lo, C. W., Border, W. L., Wechsler, S. B., Murali, C. N., Azamian, M. S., Lalani, S. R., Hinton, R. B., Garg, et al  
2023; 12 (18): e029340
- **Heterozygous TP63 pathogenic variants in isolated primary ovarian insufficiency.** *Journal of assisted reproduction and genetics*  
Vanderschelden, R. K., Rodriguez-Escriba, M., Chan, S. H., Berman, A. J., Rajkovic, A., Yatsenko, S. A.  
2023; 40 (9): 2211-2218
- **2022 Association of Professors of Human and Medical Genetics (APHMG) consensus-based update of the core competencies for undergraduate medical education in genetics and genomics.** *Genetics in medicine : official journal of the American College of Medical Genetics*  
Massingham, L. J., Nunez, S., Bernstein, J. A., Gardner, D. P., Parikh, A. S., Strovel, E. T., Quintero-Rivera, F., Association of Professors of Human and Medical Genetics Course Directors Special Interest Group Medical Education Core Curriculum Workgroup, Anderson, H., Ashfaq, M., Bernstein, J., Burke, L., Cross, C., et al  
2022
- **Pathogenic Variants in ZSWIM7 Cause Primary Ovarian Insufficiency.** *The Journal of clinical endocrinology and metabolism*  
Yatsenko, S. A., Gurbuz, F., Topaloglu, A. K., Berman, A. J., Martin, P. M., Rodrigue-Escribà, M., Qin, Y., Rajkovic, A.  
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- **Targeted whole exome sequencing and Drosophila modelling to unveil the molecular basis of primary ovarian insufficiency.** *Human reproduction (Oxford, England)*  
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- **Deletion of conserved non-coding sequences downstream from NKX2-1: A novel disease-causing mechanism for benign hereditary chorea.** *Molecular genetics & genomic medicine*  
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2021; 9 (4): e1647
- **Copy number alterations involving 59 ACMG-recommended secondary findings genes.** *Clinical genetics*  
Yatsenko, S. A., Aarabi, M., Hu, J., Surti, U., Ortiz, D., Madan-Khetarpal, S., Saller, D. N., Bellissimo, D., Rajkovic, A.  
2020; 98 (6): 577-588
- **Autism spectrum disorder in females with ARHGEF9 alterations and a random pattern of X chromosome inactivation.** *European journal of medical genetics*  
Aarabi, M., Kessler, E., Madan-Khetarpal, S., Surti, U., Bellissimo, D., Rajkovic, A., Yatsenko, S. A.  
2019; 62 (4): 239-242
- **Mutations involving the SRY-related gene SOX8 are associated with a spectrum of human reproductive anomalies.** *Human molecular genetics*  
Portnoi, M. F., Dumargne, M. C., Rojo, S., Witchel, S. F., Duncan, A. J., Eozenou, C., Bignon-Topalovic, J., Yatsenko, S. A., Rajkovic, A., Reyes-Mugica, M., Almstrup, K., Fusee, L., Srivastava, et al  
2018; 27 (7): 1228-1240
- **Chromosomal instability in women with primary ovarian insufficiency.** *Human reproduction (Oxford, England)*  
Katari, S., Aarabi, M., Kintigh, A., Mann, S., Yatsenko, S. A., Sanfilippo, J. S., Zeleznik, A. J., Rajkovic, A.  
2018; 33 (3): 531-538

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2018; 137 (2): 175-181
- **Highly heterogeneous genomic landscape of uterine leiomyomas by whole exome sequencing and genome-wide arrays.** *Fertility and sterility*  
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2017; 107 (2): 457-466.e9
- **The Genomic Landscape of PAX5, IKZF1, and CDKN2A/B Alterations in B-Cell Precursor Acute Lymphoblastic Leukemia.** *Cytogenetic and genome research*  
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2016; 150 (3-4): 242-252
- **High-resolution microarray analysis unravels complex Xq28 aberrations in patients and carriers affected by X-linked blue cone monochromacy.** *Clinical genetics*  
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- **Integration of microarray analysis into the clinical diagnosis of hematological malignancies: How much can we improve cytogenetic testing?** *Oncotarget*  
Peterson, J. F., Aggarwal, N., Smith, C. A., Gollin, S. M., Surti, U., Rajkovic, A., Swerdlow, S. H., Yatsenko, S. A.  
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- **Med12 gain-of-function mutation causes leiomyomas and genomic instability.** *The Journal of clinical investigation*  
Mittal, P., Shin, Y. H., Yatsenko, S. A., Castro, C. A., Surti, U., Rajkovic, A.  
2015; 125 (8): 3280-4
- **Application of chromosomal microarray in the evaluation of abnormal prenatal findings.** *Clinical genetics*  
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- **Human subtelomeric copy number gains suggest a DNA replication mechanism for formation: beyond breakage-fusion-bridge for telomere stabilization.** *Human genetics*  
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- **Novel 9q34.11 gene deletions encompassing combinations of four Mendelian disease genes: STXBP1, SPTAN1, ENG, and TOR1A.** *Genetics in medicine : official journal of the American College of Medical Genetics*  
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- **Noninvasive prenatal diagnosis of a fetal microdeletion syndrome.** *The New England journal of medicine*  
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- **Hormad1 mutation disrupts synaptonemal complex formation, recombination, and chromosome segregation in mammalian meiosis.** *PLoS genetics*  
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- **Identification of de novo copy number variants associated with human disorders of sexual development.** *PLoS one*  
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- **Molecular characterization of a balanced rearrangement of chromosome 12 in two siblings with Noonan syndrome.** *American journal of medical genetics. Part A*  
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- **Identification of critical regions for clinical features of distal 10q deletion syndrome.** *Clinical genetics*  
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Yatsenko, S. A., Shaw, C. A., Ou, Z., Pursley, A. N., Patel, A., Bi, W., Cheung, S. W., Lupski, J. R., Chinault, A. C., Beaudet, A. L.  
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- **Identification of chromosome abnormalities in subtelomeric regions by microarray analysis: a study of 5,380 cases.** *American journal of medical genetics. Part A*  
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