## Fedik Rahimov

List of Publications by Year in descending order

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FEDIR RAHIMON

| #  | Article  | IF   | CITATIONS |
|----|--|------|-----------|
| 1  | Concordance between gene expression in peripheral whole blood and colonic tissue in children with inflammatory bowel disease. PLoS ONE, 2019, 14, e0222952.  | 2.5  | 28        |
| 2  | Transgenic zebrafish model of DUX4 misexpression reveals a developmental role in FSHD pathogenesis.<br>Human Molecular Genetics, 2019, 28, 320-331.  | 2.9  | 14        |
| 3  | Normalizing hepcidin predicts TMPRSS6 mutation status in patients with chronic iron deficiency.<br>Blood, 2018, 132, 448-452.  | 1.4  | 16        |
| 4  | CD82 Is a Marker for Prospective Isolation of Human Muscle Satellite Cells and Is Linked to Muscular<br>Dystrophies. Cell Stem Cell, 2016, 19, 800-807.  | 11.1 | 97        |
| 5  | A missense mutation in TFRC, encoding transferrin receptor 1, causes combined immunodeficiency.<br>Nature Genetics, 2016, 48, 74-78.   | 21.4 | 219       |
| 6  | Congenital sideroblastic anemia due to mutations in the mitochondrial HSP70 homologue HSPA9.<br>Blood, 2015, 126, 2734-2738.   | 1.4  | 78        |
| 7  | Emerging preclinical animal models for FSHD. Trends in Molecular Medicine, 2015, 21, 295-306.  | 6.7  | 40        |
| 8  | An etiologic regulatory mutation in IRF6 with loss- and gain-of-function effects. Human Molecular<br>Genetics, 2014, 23, 2711-2720.  | 2.9  | 55        |
| 9  | Human skeletal muscle xenograft as a new preclinical model for muscle disorders. Human Molecular<br>Genetics, 2014, 23, 3180-3188.   | 2.9  | 48        |
| 10 | Cellular and molecular mechanisms underlying muscular dystrophy. Journal of Cell Biology, 2013, 201,<br>499-510.   | 5.2  | 203       |
| 11 | Exome sequencing identifies a novel SMCHD1 mutation in facioscapulohumeral muscular dystrophy 2.<br>Neuromuscular Disorders, 2013, 23, 975-980.  | 0.6  | 32        |
| 12 | Telomere position effect regulates DUX4 in human facioscapulohumeral muscular dystrophy. Nature<br>Structural and Molecular Biology, 2013, 20, 671-678.  | 8.2  | 95        |
| 13 | Dux4 target gene expression in mouse muscle transplanted with muscle cells from FSHD patients.<br>FASEB Journal, 2013, 27, 751.6.  | 0.5  | 0         |
| 14 | Transcriptional profiling in facioscapulohumeral muscular dystrophy to identify candidate<br>biomarkers. Proceedings of the National Academy of Sciences of the United States of America, 2012,<br>109, 16234-16239. | 7.1  | 81        |
| 15 | Facioscapulohumeral muscular dystrophy family studies of DUX4 expression: evidence for disease<br>modifiers and a quantitative model of pathogenesis. Human Molecular Genetics, 2012, 21, 4419-4430.                 | 2.9  | 150       |
| 16 | A unique library of myogenic cells from facioscapulohumeral muscular dystrophy subjects and<br>unaffected relatives: family, disease and cell function. European Journal of Human Genetics, 2012, 20,<br>404-410.    | 2.8  | 57        |
| 17 | Genetics of Nonsyndromic Orofacial Clefts. Cleft Palate-Craniofacial Journal, 2012, 49, 73-91.   | 0.9  | 212       |
| 18 | Gene expression profiling of skeletal muscles treated with a soluble activin type IIB receptor.<br>Physiological Genomics, 2011, 43, 398-407.  | 2.3  | 44        |

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|----|---|------|-----------|
| 19 | Genetic variants in <i>IRF6</i> and the risk of facial clefts: singleâ€marker and haplotypeâ€based analyses<br>in a populationâ€based caseâ€control study of facial clefts in Norway. Genetic Epidemiology, 2008, 32,<br>413-424. | 1.3  | 94        |
| 20 | Disruption of an AP-2α binding site in an IRF6 enhancer is associated with cleft lip. Nature Genetics, 2008, 40, 1341-1347.   | 21.4 | 382       |
| 21 | GLI2 mutations in four Brazilian patients: How wide is the phenotypic spectrum?. American Journal of<br>Medical Genetics, Part A, 2006, 140A, 2571-2576.  | 1.2  | 54        |
| 22 | Medical Sequencing of Candidate Genes for Nonsyndromic Cleft Lip and Palate. PLoS Genetics, 2005, 1, e64.   | 3.5  | 212       |
| 23 | Direct Sequencing of Candidate Genes for Nonsyndromic Cleft Lip and Palate. PLoS Genetics, 2005, preprint, e64.   | 3.5  | 1         |