

CONTENTS

Original Articles

INVESTIGATION OF CIRCULATING SERUM microRNA-328-3p AND microRNA-3135a EXPRESSION AS PROMISING NOVEL BIOMARKERS FOR AUTISM SPECTRUM DISORDER Popov NT, Minchev DS, Naydenov MM, Minkov IN, Vachev TI	5
 DETERMINING SPECIFIC THYROID TRANSCRIPTS IN PERIPHERAL BLOOD: A SINGLE CENTER STUDY EXPERIENCE Makazlieva T, Eftimov A, Vaskova O, Tripunoski T, Miladinova D, Risteski S, Jovanovic H, Jakovski Z	
13	
 THE FREQUENCY OF EGFR AND KRAS MUTATIONS IN THE TURKISH POPULATION WITH NON-SMALL CELL LUNG CANCER AND THEIR RESPONSE TO ERLOTINIB THERAPY Demiray A, Yaren A, Karagenç N, Bir F, Demiray AG, Karagür ER, Tokgün O, Elmas L, Akça H	
21	
 EPIGENETIC SIGNATURE OF CHRONIC MATERNAL STRESS LOAD DURING PREGNANCY MIGHT BE A POTENTIAL BIOMARKER FOR SPONTANEOUS PRETERM BIRTH Rogac M, Peterlin B.....	
27	

Case Reports

CHARACTERISTIC DIAGNOSTIC CLUES OF METATROPIC DYSPLASIA: THE LUMBOThoracic HUMPBACK WITH DUMBBELL APPEARANCE OF THE LONG BONES Gucev Z, Kalcev G, Laban N, Bozinovski Z, Popovski N, Saveski A, Daskalov B, Plaseska-Karanfilska D, Tasic V	35
 MYOTONIC DYSTROPHY-2: UNUSUAL PHENOTYPE DUE TO A SMALL CCTG-EXPANSION Finsterer J, Stöllberger C, Reining-Festa A, Loewe-Grgurin M, Gencik M.....	
39	
 NEXT GENERATION SEQUENCING IDENTIFIED A NOVEL MULTI EXON DELETION OF THE NF1 GENE IN A CHINESE PEDIGREE WITH NEUROFIBROMATOSIS TYPE 1 Yang J, An J-X, Liu X-L, Wang Z-Q, Xie G-M, Yang X-L, Xu S-J, Feng F, Ni Y	
45	
 THE ROLE OF NEXT GENERATION SEQUENCING IN THE DIFFERENTIAL DIAGNOSIS OF CAROLI'S SYNDROME Smolović B, Muhović D, Hodžić A, Bergant G, Peterlin B	
49	
 A NOVEL MUTATION IN A NEWBORN BABY LEADING TO GLYCOGEN STORAGE DISEASE TYPE IA Dorum S, Gorukmez O	
55	
 RARE CASE OF A HETEROZYGOUS MICRODELETION 9q21.11-q21.2: CLINICAL AND GENETIC CHARACTERISTICS Ivanov HY, Stoyanova V, Ivanov I, Linev A, Vazharova R, Ivanov S, Balabanski L, Toncheva D	
59	

**A NOVEL *DE NOVO* PARACENTRIC INVERSION [inv(20)(q13.1q13.3)]
ACCOMPANIED BY AN 11q14.3-q21 MICRODELETION IN A PEDIATRIC
PATIENT WITH AN INTELLECTUAL DISABILITY**

Zachaki S, Kouvidi E, Mitrakos A, Lazaros L, Pantou A, Mavrou A, Tzetis M, Manola KN 63

**PRENATAL DIAGNOSIS OF A NEW CASE: *DE NOVO* BALANCED NON-ROBERTSONIAN
TRANSLOCATION INVOLVING t(15;22)(p11.2;q11.2)**

Atli Eİ, Gurkan H, Atli E, Tozkir H, Varol GF, İnan C2 69

**A FAMILIAL CASE REPORT OF A 13;22 CHROMOSOMAL TRANSLOCATION WITH
RECURRENT INTRACYTOPLASMIC SPERM INJECTION FAILURE**

Verma S, Shah R, Bhat A, Bhat GR, Dada R, Kumar R 73