



## Rare Chromosomal Duplication Syndromes: Expanding the Phenotypic Spectrum

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**Objective:** Chromosomal duplications present diverse clinical manifestations depending on the duplicated region. While full trisomies (13, 18, 21) are well-characterized, rare forms like mosaic trisomy 8, partial trisomy 15q, and partial trisomy 17p remain less defined. This study characterizes their clinical and molecular spectrum to improve diagnostic accuracy and genetic counseling.

**Materials and Methods:** This retrospective study evaluated four patients admitted to Pamukkale University Medical Genetics, between 2020 and 2025 with suspected chromosomal anomalies. Ethical approval was obtained from the University Non-Interventional Clinical Research Ethics Committee. Patients underwent comprehensive genetic analysis including G-banded karyotyping, fluorescence in situ hybridization (FISH), and chromosomal microarray analysis (CMA). Clinical data were retrieved from medical records and analyzed alongside cytogenetic findings.

**Results:** The study cohort consisted of four female patients. Cytogenetic analysis identified mosaic trisomy 8 in two patients (mosaicism rates 48% and 54%), partial trisomy 15q secondary to an unbalanced translocation in one patient, and partial trisomy 17p in one patient. Common clinical features included hypotonia (100%), developmental delay (75%), and craniofacial dysmorphism (100%). Novel findings such as leukocoria were observed in mosaic trisomy 8 cases. CMA successfully refined breakpoints in microduplication syndromes where karyotyping was insufficient.

**Conclusion:** Rare chromosomal duplications exhibit significant clinical heterogeneity. This study underscores the necessity of integrating conventional karyotyping with high-resolution techniques like CMA for precise diagnosis. Our findings expand the known phenotypic spectrum of these syndromes and highlight the critical role of multidisciplinary management and family-based genetic counseling in improving patient outcomes.

**Key Words:** Mosaic trisomy 8, partial trisomy 15q, partial trisomy 17p, chromosomal microarray, phenotype genotype spectrum

### Nadir Kromozomal Duplikasyon Sendromları: Fenotipik Spektrumun Genişletilmesi

**Amaç:** Kromozomal duplikasyonlar, duplike olan bölgeye bağlı olarak çeşitli klinik bulgular gösterir. Tam trizomiler (13, 18, 21) iyi tanımlanmış olsa da mozaik trizomi 8, kısmi trizomi 15q ve kısmi trizomi 17p gibi nadir formlar yeterince tanımlanmamıştır. Bu çalışma, tanınal doğruluğu ve genetik danışmanlığı iyileştirmek amacıyla bunların klinik ve moleküler spektrumunu karakterize etmeyi amaçlamaktadır.

**Gereç ve Yöntem:** Bu retrospektif çalışmada, 2020-2025 yılları arasında şüpheli kromozomal anomalilerle Pamukkale Üniversitesi Tıbbi Genetik'e başvuran dört hasta değerlendirilmiştir. Üniversite Girişimsel Olmayan Klinik Araştırmalar Etik Kurulu'ndan etik onay alınmıştır. Hastalar G-bantlı karyotipleme, floresan in situ hibridizasyon (FISH) ve kromozomal mikroarray analizi (CMA) dahil kapsamlı genetik analizlere tabi tutulmuştur. Klinik veriler tıbbi kayıtlardan alınmış ve sitogenetik bulgularla birlikte analiz edilmiştir.

**Bulgular:** Çalışma kohortu dört kız hastadan oluşmuştur. Sitogenetik analizde iki hastada mozaik trizomi 8 (mozaik oranları %48 ve %54), bir hastada dengesiz translokasyona bağlı kısmi trizomi 15q ve bir hastada kısmi trizomi 17p saptanmıştır. Yaygın klinik bulgular arasında hipotoni (%100), gelişimsel gerilik (%75) ve kraniyofasiyal dismorfizm (%100) yer almıştır. Mozaik trizomi 8 olgularında lökokori gibi yeni bulgular gözlenmiştir. CMA, karyotipleminin yetersiz kaldığı mikroduplikasyon sendromlarında kırılma noktalarını başarıyla hassaslaştırmıştır.

**Sonuç:** Nadir kromozomal duplikasyonlar önemli klinik heterojenite sergilemektedir. Bu çalışma, kesin tanı için konvansiyonel karyotipleminin CMA gibi yüksek çözünürlüklü tekniklerle entegrasyonunun gerekliliğini vurgulamaktadır. Bulgularımız bu sendromların bilinen fenotipik spektrumunu genişletmekte ve hasta sonuçlarının iyileştirilmesinde multidisipliner yönetim ve aile temelli genetik danışmanlığın kritik rolünü ortaya koymaktadır.

**Anahtar Kelimeler:** Mozaik trizomi 8, kısmi trizomi 15q, kısmi trizomi 17p, duplikasyon sendromları, fenotip spektrumu

### Introduction

Chromosomal duplications involve the formation of additional copies of a specific chromosomal region, resulting in varying gene copy numbers within that region. Duplications can impact the phenotype by altering gene dosage, as the amount of protein synthesized is often proportional to the number of gene copies. Consequently, extra genes can lead to an excess of proteins (1).

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While full trisomies of chromosomes 13, 18, and 21 are the most commonly observed autosomal duplications at birth, other trisomies such as mosaic trisomy 8, trisomy 15q, and trisomy 17p have also been identified and are compatible with life.

Among these, mosaic trisomy 8 (T8MS), also known as Warkany Syndrome, is a rare chromosomal disorder characterized by the presence of an extra chromosome 8 in some cells. It exhibits distinct clinical features and an estimated incidence of 1 in 30,000 live births, stands out as a well-known syndrome associated with a range of developmental abnormalities. To date, approximately 121 cases have been reported in the literature, with a male-to-female ratio of 5:1. Most infants with T8MS have a normal life expectancy and birth weight. T8MS occurs due to postzygotic mitotic dissociation and manifests as a clinically heterogeneous syndrome (2, 3). Common features associated with T8MS include dysmorphism, developmental delay, intellectual disability, cardiac and renal anomalies, agenesis of the corpus callosum, spinal deformity, scoliosis, hemivertebrae, contractures of the fingers and toes, and moderate mental retardation (4, 5).

Duplication of chromosome 15q (dup15q), also known as trisomy 15q, is another rare chromosomal disorder that typically arises from unbalanced translocations resulting in 15q duplication. It was first described in 1974 by Fujimoto et al. (6), and at least 50 cases have been reported to date (7). Although the breakpoints in 15q duplications may vary, the general phenotype remains similar. Dup15q is characterized by craniofacial malformations, such as an elongated face, down slanting palpebral fissures, micrognathia, a long philtrum, a broad nasal bridge, a high-arched palate, short neck, malformations of the fingers and toes, skeletal malformations, genital abnormalities, and, in some cases, cardiac defects. Additional abnormalities typically include post- and pre-natal overgrowth, mild to profound intellectual disability, hypotonia, and motor delays (8, 9). The severity of symptoms varies depending on the location and length of the duplicated portion of chromosome 15q (10).

Duplication of chromosome 17p is a rare chromosomal abnormality resulting in distinct clinical features, such as dysmorphic facial appearances (ptosis, malformed ears, and abnormality of the iris micrognathia, high-arched palate), prenatal and postnatal growth retardation, developmental delay, hypotonia, digital abnormalities, behavioral difficulties such as hyperactivity and autistic features, speech and language delay, intellectual disability, feeding difficulties, and congenital heart defects. Duplication 17p syndromes are classified into four groups based on the duplicated region (11): Group 1 involves an extra copy of short arm band 17p11.2 and is known as Potocki-Lupski syndrome (PTLS). Group 2 involves duplication of band 17p11.2 and/or 17p12, usually showing features of PTLS with additional features (12, 13). Group 3 involves an extra copy of short arm band 17p13 and is known as 17p13 microduplication syndrome (14). Finally, group 4, known as trisomy 17p, involves a complete duplication of the short arm of 17p and is extremely rare (11).

This study aims to characterize the clinical and cytogenetic spectrum of four patients with rare chromosomal duplications involving mosaic trisomy 8, partial trisomy 15q, and partial trisomy 17p. By integrating conventional karyotyping with high-resolution molecular techniques, we seek to expand the known phenotypic spectrum and highlight the diagnostic challenges associated with these rare anomalies.

## Materials and Methods

**Research and Publication Ethics:** This study was approved by the Pamukkale University Non-Interventional Clinical Research Ethics Committee (Approval date and number: 13.08.2025-E.735017). Written informed consent was obtained from the legal guardians of all patients prior to the collection of blood samples.

**Study Design and Patient Selection:** This retrospective study evaluated four patients admitted to Pamukkale University, Department of Medical Genetics, between 2020 and 2025 with suspected chromosomal anomalies. Inclusion criteria were: (1) presence of dysmorphic features and/or developmental delay, and (2) confirmed chromosomal duplications via G-banded karyotyping and/or chromosomal microarray analysis. Clinical data, including demographic characteristics, physical examination findings, and imaging results, were retrospectively retrieved from electronic medical records and analyzed alongside cytogenetic findings.

**Cytogenetic Analysis:** Peripheral venous blood samples were collected in heparinized tubes for chromosomal analysis. Lymphocytes were cultured in RPMI-1640 medium supplemented with phytohemagglutinin (PHA) and incubated at 37 °C for 72 hours. Colcemid was added 45 minutes prior to harvesting. Cultured cells underwent hypotonic treatment followed by fixation with Carnoy's fixative. Metaphase spreads were prepared, aged, and analyzed after G-banding. A minimum of 20 metaphase spreads were evaluated per patient under a light microscope.

**Fluorescence in situ Hybridization (FISH):** FISH analysis was performed to investigate specific chromosomal abnormalities, following the manufacturer's protocols. Commercially available DNA probes targeting chromosome-specific regions (Vysis probes) were used. Hybridized slides were analyzed and imaged using the ISIS imaging system (MetaSystems).

**Chromosomal Microarray Analysis:** Chromosomal microarray analysis was conducted using the HumanCytoSNP-12 DNA Analysis BeadChip v2.1 kit (Illumina, San Diego, CA, USA), containing approximately 299,140 SNP markers. Data were analyzed using GenomeStudio Data Analysis Software v2.0.4, with reference to the human genome build GRCh37/hg19. Interpretation was performed in accordance with the guidelines of the International Standards for Cytogenomic Arrays (ISCA) Consortium and the American College of Medical Genetics and Genomics (ACMG) (15-16).

**Table 1.** Clinical features and cytogenetic findings of the cases

Patient	Case 1	Case 2	Case 3	Case 4
<b>Karyotype</b>	<b>mos47,XX,+8[24]/46,XX[26]</b>	<b>mos47,XX,+8[54]/46,XX[46]</b>	<b>46,XX,der(9) t(9;15)(p22.1;q23)pat</b>	<b>46,XX,dup(17)(p13.1p11.1)</b>
Gender	F	F	F	F
Birth weight	3250	3200	2875 (25-50p)	3250
Age at diagnosis	2y	1y	Newborn	1y
Weight (kg)	10 (3p)	6150gr	2730 (10-25p)	Na
Height (cm)	84 (25-50p)	72	52 (50-75p)	Na
OFD (cm)	47 (75-90p)	43.5	33 (5-10p)	Na
<b>Facial dysmorphism</b>				
Asymmetric face	+	+	+	-
Curly hair	+	+	-	-
Leukokoria	+	-	-	-
Prominent nasal bridge	-	+	+	-
Depressed nasal root	-	+	+	+
Broad nasal bridge	+	-	+	-
Deep set eyes	-	-	+	-
Preauricular sinus	-	-	-	-
Posteriorly rotated ears	-	-	-	-
Large ears	-	-	+	-
Long philtrum	-	+	+	-
Short philtrum	-	-	-	-
Micrognathia	-	+	-	+
Full lips	-	-	-	-
High palate	-	+	+	+
Cleft palate	-	+	-	-
Broad nasal tip	+	+	-	+
Prominent chin	-	-	+	-
<b>Gastrointestinal system findings</b>				
Gastroesophageal reflux	+	-	-	+
Feeding difficulties	-	+	+	+
Constipation	-	+	+	-
<b>Skeletal system findings</b>				
Pes varus	-	-	-	-
Kyphoscoliosis	-	+	-	-
Joint laxity	+	-	-	+
Long slenderic fingers	-	+	+	-
Small feet	-	+	-	-
Deep plantar creases	+	+	-	-
Overlapping toes	-	+	-	-
<b>Neurologic system findings</b>				
Developmental delay	+	+	na	+
Hypotonia	+	+	+	+
Spasticity	-	-	-	-
Purposeful hand use	+	-	-	-
Early death	-	-	-	-
<b>Cranial MRI findings</b>				
Agenesis of corpus callosum	-	+	na	-
Colpocephaly	+	-	-	-
Hypoplasia of corpus callosum	+	-	na	-

F: Female; na: Not available



**Figure 1.** Facial Dysmorphisms in patients (eyes obscured for privacy). **Case 1.** Mosaic Trisomy 8 syndrome, in a 2.5 years old girl. Note an asymmetric face, curly hair, leukocoria, broad nasal bridge, broad nasal tip. **Case 2.** Mosaic Trisomy 8 syndrome in a 1-year-old girl. **Case 3.** A newborn with duplication chromosome 15q is shown. **Case 4.** Duplication 17p syndrome in a 1-year old girl.

## Results

**Demographic and Clinical Characteristics:** The study cohort consisted of four female patients aged between the newborn period and 2.5 years. All patients were referred to the Department of Medical Genetics due to dysmorphic features and/or developmental delays. Two patients were diagnosed with mosaic trisomy 8, one with partial trisomy 15q secondary to an unbalanced translocation, and one with partial trisomy 17p.

Birth weights ranged from 2875 g to 3250 g. Postnatal growth parameters indicated growth retardation in 50% of the patients (weight <3rd or 10th percentile). Clinical evaluation revealed significant phenotypic heterogeneity. Hypotonia was observed in all patients (100%), while developmental delay was documented in 75% of the cohort (one patient was a newborn and could not be assessed for neurodevelopment). Feeding difficulties were present in 75% of the patients, accompanied by gastroesophageal reflux in 50% and constipation in 50%.

Craniofacial dysmorphisms were prominent across the cohort. Facial asymmetry was noted in 75% of the patients. Nasal anomalies were frequent, including a depressed nasal root (75%), broad nasal bridge (50%), and broad nasal tip (75%). High-arched palate was observed in 75% of the patients, while micrognathia was present in 50%. Skeletal anomalies included joint laxity

(50%), deep plantar creases (50%), and overlapping toes (25%). Neuroimaging, performed in two patients, revealed midline brain defects; one patient exhibited agenesis of the corpus callosum, while another showed corpus callosum hypoplasia and colpocephaly. Ocular anomalies were noted in one patient (leukocoria). Detailed clinical features are summarized in Table 1.

**Cytogenetic and Molecular Findings:** Cytogenetic analysis confirmed chromosomal duplications in all patients using G-banded karyotyping, supplemented by FISH and Chromosomal Microarray Analysis (CMA) where indicated (Table 2).

In the two patients with mosaic trisomy 8, karyotyping revealed mosaicism rates of 48% and 54% in peripheral blood lymphocytes (mos47,XX,+8[24]/46,XX[26] and mos47,XX,+8[54]/46,XX[46], respectively). No further molecular analysis was required for these cases.

The patient with partial trisomy 15q presented with an unbalanced derivative chromosome 9. Karyotyping identified the structure as 46,XX,der(9)t(9;15)(p22.1;q23), indicating partial trisomy of 15q23-qter and partial monosomy of 9p22.1-pter. FISH analysis confirmed the unbalanced translocation using chromosome-specific probes. Parental karyotyping revealed a normal maternal karyotype and a balanced reciprocal translocation t(9;15) in the father.

The patient with partial trisomy 17p showed a duplication of the short arm of chromosome 17 via conventional karyotyping (46,XX,dup(17)(p13.1p11.1)). Subsequent CMA refined the breakpoint and size of the

duplication, identifying a 9.6 Mb gain in the 17p13.1-p11.2 region (arr[GRCh37]17p13.1p11.2(9,262,182\_18,905,375)x3).

**Table 2.** Chromosome, FISH, and microarray results of all patients

Patient	Karyotype	FISH	aCGH
Case 1	mos47,XX,+8[24]/46,XX[26]	-	-
Case 2	mos47,XX,+8[54]/46,XX[46]	-	-
Case 3	46,XX,der(9)t(9;15)(p22.1;q23)pat	46,XX,der(9)t(9;15)(p22;q23)pat.ish.der(9)t(9;15)(305J7T7-, D15S936+, CDKN2A+, PML-)	-
Case 4	46,XX,dup(17)(p13.1p11.1)	-	arr[GRCh37]17p13.1p11.2(9262182-18905375)x3

## Discussion

Chromosomal duplication syndromes represent a complex and heterogeneous group of disorders with variable phenotypic expression. Our case series of four patients with rare duplications mosaic trisomy 8, partial trisomy 15q, and partial trisomy 17p provides further insights into the clinical and molecular spectrum of these conditions while highlighting key diagnostic and management challenges.

**Mosaic Trisomy 8: Expanding the Phenotypic Variability:** The patients with mosaic trisomy 8 (T8MS) in this cohort exhibited classic features such as facial asymmetry, joint laxity, and developmental delay, consistent with prior reports (2, 3). Notably, the degree of mosaicism (48% and 54%) did not directly correlate with clinical severity, reinforcing the notion that the tissue-specific distribution of trisomic cells may play a more critical role than the overall mosaicism percentage detected in peripheral blood (17). Beyond these classic features, ocular anomalies such as leukocoria and corpus callosum hypoplasia were observed in one patient, findings that significantly expand the known T8MS phenotype. While ocular anomalies like corneal opacities, strabismus, and colobomas have been previously described in T8MS, leukocoria is a distinct clinical sign often indicative of underlying pathologies such as cataracts or retinal dysgenesis. Furthermore, the concurrent presence of corpus callosum hypoplasia suggests a disruption in early midline developmental processes. We hypothesize that the dosage effect of developmental regulators located on chromosome 8, such as CHD7 (8q12.2) or SOX17 (8q11.23), combined with tissue-specific mosaicism, contributes to these rare CNS and ocular manifestations (18). Consequently, these findings underscore the necessity of cranial imaging and detailed ophthalmologic evaluation in patients with T8MS, even in the absence of severe neurological deficits. Finally, the absence of major cardiac or renal malformations in this cohort contrasts with some previous studies (4), further suggesting that phenotypic variability is heavily influenced by the timing of postzygotic nondisjunction and the specific cell lineages affected.

## Partial Trisomy 15q: The Impact of Concurrent

**Genomic Imbalance:** One patient presented with an unbalanced translocation resulting in partial trisomy 15q23-qter and partial monosomy 9p22.1-pter. While the dysmorphic features (remarkable nasal root, micrognathia) and feeding difficulties align with previous reports of dup15q (6, 8), the observed growth retardation is more characteristic of monosomy 9q than isolated 15q duplication. This underscores the importance of comprehensive cytogenetic analysis in cases of complex rearrangements, as concomitant genomic imbalances can significantly modify the phenotype. Parental studies revealed a paternal balanced translocation, emphasizing the need for familial testing to assess recurrence risks. Notably, the absence of severe intellectual disability in this infant contrasts with earlier descriptions of dup15q (10), suggesting that distal 15q duplications may have a milder neurocognitive impact compared to proximal duplications involving the Prader-Willi/Angelman critical region.

## Partial Trisomy 17p: Refining the Genotype-Phenotype Correlation:

The 9.6 Mb duplication in 17p13.1-p11.2 observed in one patient falls within the Potocki-Lupski syndrome (PTLS) spectrum, yet the phenotype lacked some classic features such as hyperactive behavior and congenital heart defects (11, 13). This variability may reflect differences in the exact breakpoints or the influence of modifying genetic or epigenetic factors. The presence of RAI1 in the duplicated region likely explains the shared core features (e.g., developmental delay, hypotonia), while the absence of certain PTLS-associated traits suggests that additional genes or regulatory elements outside this interval may contribute to the full syndrome (19). Chromosomal microarray analysis was indispensable in this case, as conventional karyotyping could not precisely define the duplication's boundaries, demonstrating the superiority of high-resolution techniques in diagnosing microduplication syndromes.

In conclusion, the findings of this study underscore the significant clinical heterogeneity of rare chromosomal duplications and highlight the diagnostic value of combining conventional karyotyping, FISH, and

chromosomal microarray analysis to characterize complex chromosomal rearrangements, especially in cases involving mosaicism or submicroscopic duplications. The observed phenotypic variability emphasizes the need for individualized management strategies.

In this cohort, precise genetic diagnosis directly influenced clinical decision-making: identification of a Potocki–Lupski syndrome–related duplication prompted early targeted interventions, while detection of a parental balanced translocation provided critical information for recurrence risk counseling. Overall, this work adds to the growing body of literature on rare chromosomal duplication syndromes and emphasizes the importance of multidisciplinary collaboration among geneticists, clinicians, and researchers to improve diagnosis,

management, and long-term outcomes for affected individuals.

Further studies incorporating advanced genomic technologies (e.g., long-read sequencing, single-cell analysis) are needed to elucidate the mechanisms underlying phenotypic variability in these disorders.

Additionally, longitudinal follow-up of patients with well-characterized duplications will help define natural history and optimize management strategies.

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**Data Availability Statement:** The data are available on request from the corresponding author.

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