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CLINICAL reports

# Oral findings in Trisomy 8 mosaicism

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## Abstract

*Trisomy 8 is a genetic dysfunction with wide-ranging, clinical defects including prominent dysmorphism affecting the head and neck. Over seventy cases have been reported in the literature since 1961; however, this paper is the first complete report of oral and dental findings in an eleven-month-old patient afflicted with the mosaic form of Trisomy 8.*

## Introduction

Numerical abnormalities in chromosomal conditions are chiefly a result of nondisjunction, i.e. the failure of paired or mated chromosomes to disjoin during the anaphase period of mitosis or meiosis. Disjunctional errors most commonly change the normal state of diploidy to a condition of monosomy or trisomy. If nondisjunction occurs during the development of the unfertilized ovum, then following fertilization, every cell of the embryo will be defective. If nondisjunction occurs early in the development of the fertilized ovum, two or more cell lines may develop, each having different chromosomal makeup. This condition of differing karyotypes or chromosomal patterns in a single individual is termed mixoploidy or mosaicism. The proportion of normal and abnormal cells vary from tissue to tissue. The mosaic individual may defy detection because repeated tissue samples from different sites may appear normal. It is also pertinent to a discussion of a genetic anomaly to include a brief review of chromosomal nomenclature. The normal male karyotype consists of 22 pairs of numbered autosomes arranged in groups A to G, plus an X and a Y sex chromosome. The female karyotype has two X chromosomes. A normal karyotype would thus be expressed as 46, XY or XX, and a trisomic condition expressed as 47, XY or XX, with a chromosome identifier, i.e. 47, XY, 8+.

With the advent of Quinacrine fluorescent and Giemsa staining techniques, it has become possible to clearly identify chromosomes not only by morphology but by the specificity of light and dark band patterns.[1] The use of Q and G banding techniques in instances of C group trisomy anomalies, has shown most cases to involve chromosomes 8, 9, and 10.[2] Trisomy 9 has been reported as the most disabling condition of the group with microcephaly and severe psychomotor retardation being prominent.[3,4,5] The diverse clinical presentations of the C group trisomies are explained by the presence of six chromosomes in the group, the mosaic condition generally being less serious than full trisomy, and the clinical picture varying with the chromosome involved and the proportion of abnormal cells in various tissues. Homogenous trisomy C group expression was formerly thought to be incompatible with life until the viable delivery of an infant with full trisomy involving chromosome 10, 11, or 12.[6] Mino *et al.* reported five cases of full Trisomy 8, but cautioned that mosaicism cannot be ruled out when only one tissue is studied.[7]

Over 70 cases of Trisomy 8 have been reported since the first report in 1961.[8] Reviews with tabulations of signs and symptoms are presented by Kakati *et al.*,[9] Fineman *et al.*,[10] and Riccardi.[11] Prominent findings common to most patients are moderate mental retardation, osseous abnormalities, restricted articular function, malformed, low-set ears, broad, upturned nose, long, slender trunk, cardiac and urogenital deformities, absent patellae, micrognathia, and palatal deformities. Although general physical findings in Trisomy 8 have been extensively documented, the incidence and description of oral anomalies has not been remarkable perhaps due to the largely medical orientation of the observers. Palatal defects are the most common oral findings, described usually as high arch-

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ing often concurrent with partial clefting. Micro or retrognathia with prominent everted lips is also commonly reported. Unusual oral findings reported are broad lingual frenum,[12,13] short teeth,[14] "fishmouth",[15] premature eruption of teeth,[16] and ridging of the alveolar gingiva.[13]

## Report of a Case

An 8.5 pound white male was born at 36 weeks gestation to a 22-year-old nullipara mother. The pregnancy was unremarkable except for mild eclampsia at week 32. Significant findings at delivery were a generalized hypotonia with depressed sucking reflex, low-set ears, large flat nose, webbed neck, pectus excavatum, widely spaced nipples, mongoloid obliquity of the eyes, and bilateral genital hydrocoele. The extremities were cyanotic. The hands and feet appeared short and wide with prominent skin fold patterns, overriding fifth toes and syndactly of third and fourth toes. Radiographic findings showed multiple rib and vertebral anomalies, missing patellae, and an increase in heart size and pulmonary vasculature. Subsequent diagnosis was made of a large ventricular septal defect and uretero-pelvic junction obstruction with mild hydroephrosis. Oral findings noted by the attending physician were a high arched palate and tongue fasciculations.

The ventricular septal defect later closed spontaneously and surgical correction of the inguinal hernia and urinary obstruction was made at five months. Developmental assessment at seven months showed delayed development in areas of gross motor adaptation and language.

The initial clinical diagnosis of a genetic abnormality was confirmed by definitive cytogenetic banding studies with the designation of Trisomy 8, mosaic form (Figure 1).

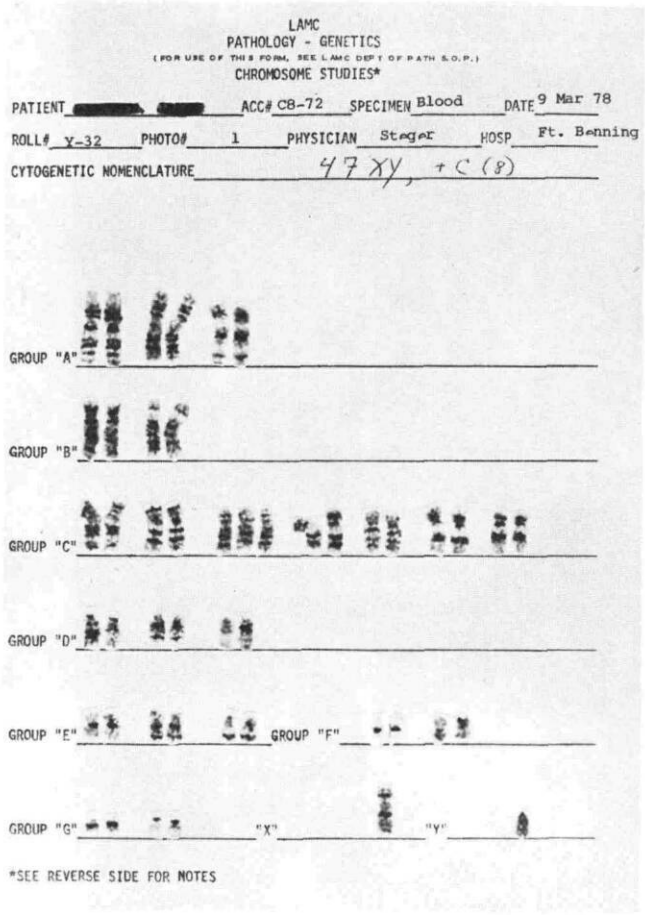


**Figure 1.** Chromosome grouping of the subject showing a Trisomy 8 mosaicism pattern.

## Dental Findings

At age 11 months, the child was referred to the Pedodontic Service for evaluation because of the mother's concern over the prematurely erupting and unsightly teeth. Dental examination disclosed the following findings:

1. Broad palate with normal height, and wide alveolar ridges.
2. Premature eruption of primary teeth. All primary teeth, except the second molars, were partially or fully erupted. The primary central incisors had erupted at eight weeks of age. The teeth were bucally positioned with hypoplastic brownish enamel suggesting incomplete matrix formation and calcification.
3. A shallow groove or indentation on the crest of the maxillary alveolar ridge extending the length of the buccal segments. (Figure 2).
4. Radiographs of the head taken at birth show the posterior deciduous teeth developing in a very superficial location on the crest of the bony alveolus. The dental follicles were positioned in shallow depressions with alveolar bone encompassing only the most radicular portions.
5. Procumbent everted fleshy lips, open mouth habitus with mouth breathing and poor tonus of tongue and perioral musculature (Figure 3).

Repeat examination of the child at 12 and 14 months showed a pattern of accelerated tooth eruption with enamel hypoplasia on the maxillary primary incisors, and first primary molar cusp tips and a chalky appearance of the enamel of other teeth.

## Discussion

Due to the preponderance of the mosaic condition

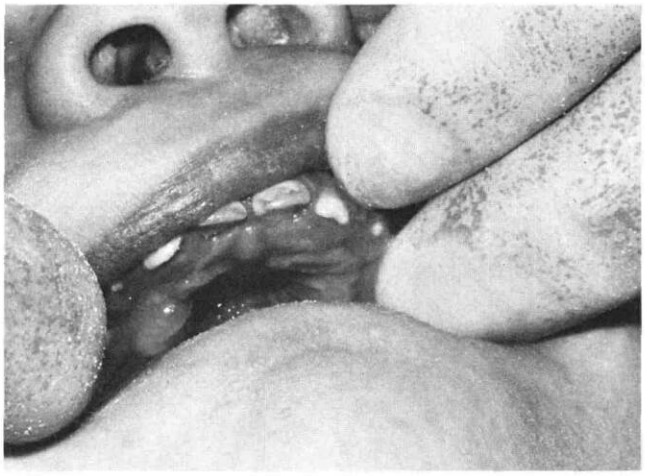

**Figure 2.** Oral findings include shallow indentation on the crest of the maxillary alveolar ridge, accelerated eruption pattern and enamel hypoplasia of the incisors and chalky appearance of the remaining teeth.

in the reported cases of Trisomy 8, the spectrum of clinical features found is quite broad. The dilution of the full expression state of Trisomy 8 by the mosaic condition not only causes phenotypic variation among patients, but also asymmetrical stigmata in the same patient.[17] The clinical features of this patient are shared by many genetic disorders, and although the pattern of the findings is suggestive of Trisomy 8, some authors do not feel that a syndrome is as yet identifiable.[9,18] One finding, however, has been reported as pathognomonic for Trisomy 8, that of deep longitudinal furrows on the soles[19,20,21] (Figure 4). The plantar furrows have been present in this patient since birth, however, they are not usually reported in older Trisomy 8 patients and may disappear with advancing age.

This case report is the first to concentrate on oral findings in Trisomy 8, and some interpretive comment is necessary. The everted lips found in this patient are similar to those appreciated in most published photographs. In this case, the lip positioning and contour were interpreted as due to incompetence of the perioral musculature. One of the most prominent features of the Trisomy 8 condition is a variable degree of poor neuromuscular tone which affected this patient's hand-eye coordination, head positioning, locomotion, as well as lip, tongue and myognathic control.

The presence of poor muscle control, mouth breathing, everted lips, and drooling habitus of Trisomy 8 patients may be responsible for the "micrognathia" noted by most observers. An apparent micro or retrognathia caused by facial posture would explain the fact that by 11 months, our patient did not have the micrognathia noted at birth. The poor perioral tonus

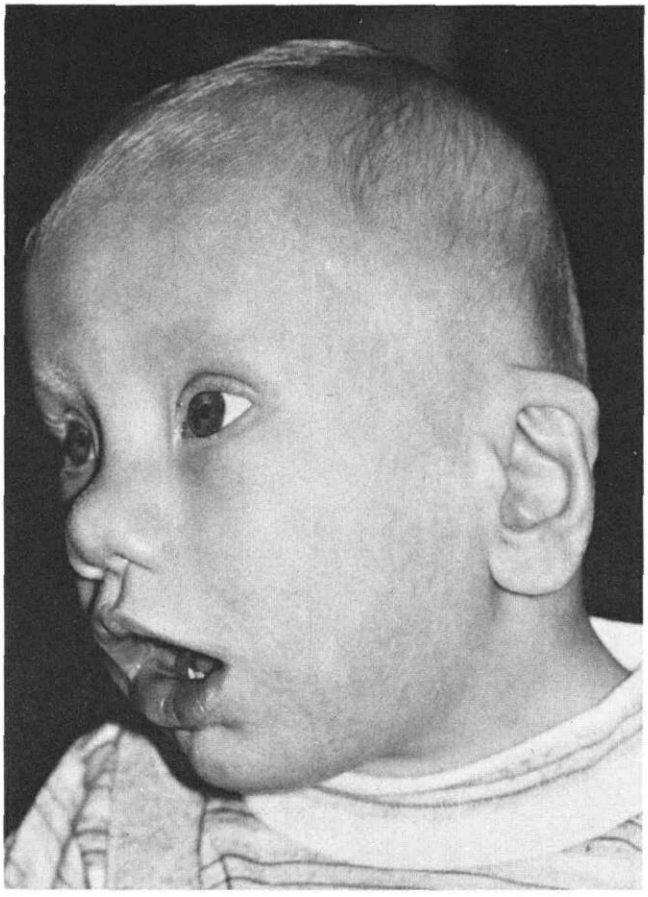

**Figure 3.** Facial characteristics include procumbent everted fleshy lips, poor tonus of tongue and perioral musculature and open mouth habitus.

would also explain the buccal drifting observed in the erupting teeth, and perhaps explain Riccardi's observation that in a 4-year-old male, the "mandibular teeth projected beyond the maxillary teeth."[15]

The alveolar grooving, and premature eruption found in this patient appear similar to instances reported by others,[13,16] and may be linked to the radiographic evidence of a defect in tooth bud positioning. If the tooth follicle develops in too shallow a position within the alveolus, the crestal bone could remain discontinuous with resultant grooving of the gingiva. Similarly, shallow positioning of developing follicles could lead to premature eruption, a mechanism used to explain the eruption of natal teeth.[23]

The enamel defects seen in our patient have not been reported previously although examination of published photographs of a five-year-old Trisomy 8 show what appears to be enamel defects on the primary mandibular lateral incisors.[15]

The condition of high arched palate so common to other cases was noted at birth by the attending physician, however, dental examination at 11 months

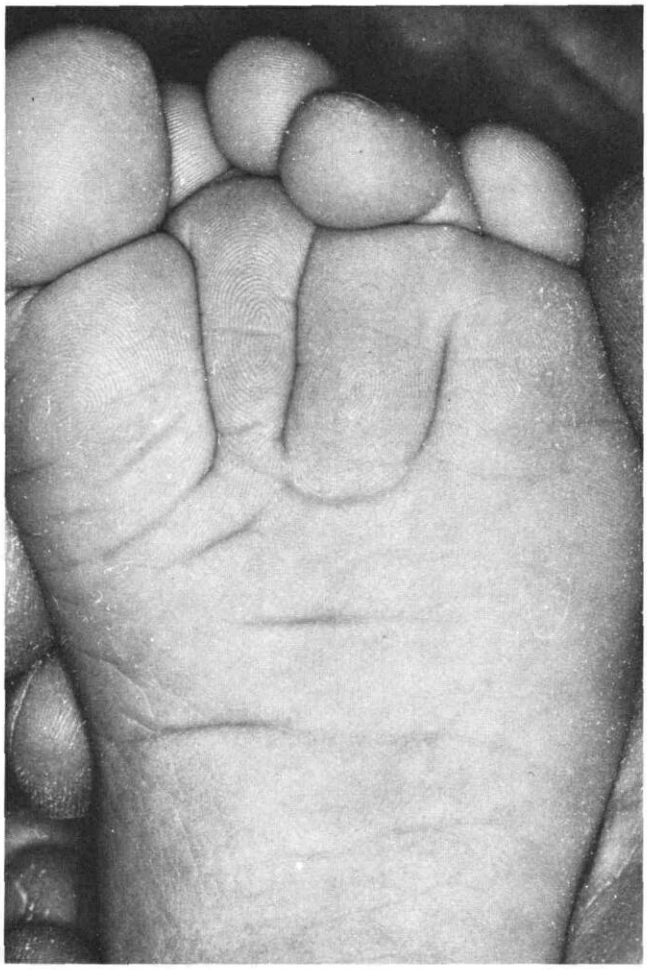

**Figure 4.** The deep longitudinal furrows on the sole is pathognomonic for Trisomy 8.

found a shallow palate but with broad alveolar ridges. It must be realized that the description of high palate is a subjective one and easily influenced by the impositions of wide alveolar ridges on the palate space leading to an impression of palatal narrowing and vaulting.

Several prognostic comments may be made relative to the expected physical and mental development of this patient. Gross and fine motor adaptation will likely remain poor. Mental retardation is generally milder than in other trisomy states and slow progress may be expected in social and language skills.[7] Our patient exhibited mild scoliosis and severe bowing of the legs necessitating orthopedic treatment at one year of age. Skeletal dysplasias and restricted articular function are reported in most cases of Trisomy 8, and more orthopedic problems can be expected in this child. At least fourteen cases of Trisomy 8 have demonstrated hematopoetic dysfunction, and one adult male developed aplastic anemia progressing to acute leukemia.[24]

This case report has been designed to direct closer attention to the oral and dental anomalies that may affect the Trisomy 8 patient. It is hoped that new case reports of this interesting entity will discover and define oral findings that may aid the clinician in recognizing the phenotype, and prompt early correction of cardiac and urinary defects.

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