

Abnormalities in tooth morphology, structure and dentition in two children with chromosome aberrations. Translocation trisomy 13 and trisomy 21

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ABSTRACT

Purpose: Dental malformations due to chromosomal trisomies are rarely described and need an intensive cooperation between pediatricians, orthodonticians and human geneticists to enable the collection of data and to extend the investigations on specific parameters of the teeth.

Results: Here we present tooth studies of two children with trisomies 13 (Patau-syndrome) and 21 (Down-syndrome): the dentition, the tooth morphology and the structure as well as the composition were investigated over a period of six years. Both male patients showed a delayed and abnormal dentition. Morphologic and structural changes compared to the general population were also detectable; whereas, the composition of the teeth was unchanged in enamel, dentin, and the border between them.

Conclusions: The abnormalities in all parameters investigated were more pronounced in the patient with Patau-syndrome than in the child with Down-syndrome.

Key words: Patau-syndrome, Down-syndrome, tooth abnormalities

INTRODUCTION

Dental malformations often escape investigation in children with chromosome syndromes caused by autosomal trisomies because of the significantly reduced life expectancy of the majority of patients. An intensive cooperation for years is required between pediatricians, orthodonticians and human geneticists to enable the collection of data and to extend the investigations on specific parameters of the teeth.

The majority of investigations in patients with chromosome syndromes are carried out in individuals with Down-syndrome, because this trisomy is frequent (1:650 live births) and the life expectancy is almost normal. The peculiarities in dentition and tooth structure are usually analyzed in the permanent teeth. The findings are then compared to single cases of chromosome disorders where patients with an usually significantly reduced life expectancy show long term survival, such as trisomy 13 or 18.

Abnormalities in tooth development are caused by complex disturbances during early embryonic development. As a basic abnormality, a change of the length of the mitotic cell cycle is discussed. This is a general symptom in different autosomal chromosome aberrations and therefore of special interest. Furthermore, an abnormal blood supply in the jaw of the embryo can hamper the tissue growth and thus lead to partial secondary degeneration of odontoblasts.

Finally a severe neurological disturbance is discussed as the cause of abnormalities in dentition. But further investigations are required to get sufficient information on this complex field of development.

In two children with autosomal chromosome disorders (trisomy 21 and translocation trisomy 13), we performed a longitudinal study to analyze the development of the deciduous teeth. The following parameters were investigated in detail: dentition, abnormalities of the tooth morphology, structure and composition.

Figure 1. Patient 1 with Patau-syndrome at birth. Tooth with sack (71). It got lost during the first month of life.



Figure 2. Patient 1 at the age of three years. Duplication of the tooth in position 52, partial retention (cutting teeth) of 53, 62 and 72. The tooth bud (71) present at birth got lost.



CASE PRESENTATIONS

Patient 1:

The boy showed a spectrum of symptoms characteristic for patients with Patau-syndrome. The development of the child could be followed up until the age of almost five years. The chromosome analysis from lymphocyte culture revealed an unbalanced Robertsonian translocation 13/14 (karyotype: 46,XY,der(14),t(13;14)(p11.2;q11) QFQ).

Tooth analyses:

The dentition was delayed and the eruption altered compared to normal children. The teeth showed morphologic changes (Fig. 1, Tab. 1). At birth, the patient already had one tooth (71 with sack) which has lost during the first year of life. One further incisive (51) developed as a double structure (Fig. 2). The structure of the teeth showed changes in the relation of enamel, dentin and pulpa. The number of dental canaliculi was reduced. They had a partially irregular arrangement (Fig. 3, Fig. 4). To further analyze the composition of the teeth the concentrations of the following elements were determined: P, F, Cl, K, Ca, Mg, Fe, Sr, S and Si. Two teeth of the patient were

Table 1. Retardation of dentition and abnormalities of shape and number (especially of the upper incisivae).

Age	Patient	Control group
Birth	71 with sack	-
20 month	51*, 52	51, 52, 53, 54
	61+61, 62	61, 62, 63, 64
	71~, 0	71, 72, 73, 74
	81, 82, 83, 84	81, 82, 83, 84
36 month	51*, 52, 53	51, 52, 53, 54, 55
	61+61, 62, 63	61, 62, 63, 64, 65
	0, 72, 73	71, 72, 73, 74, 75
	81, 82, 83, 84	81, 82, 83, 84, 85
48 month	51*, 52, 53, 54, (55)	51, 52, 53, 54, 55
	61+61, 62, 63, 64, 65	61, 62, 63, 64, 65
	0, 72, 73, 74	71, 72, 73, 74, 75
	81, 82, 83, 84	81, 82, 83, 84, 85
56 month	51*, 52, 53, 54, 55	51, 52, 53, 54, 55
	61+61, 62, 63, 64, 65	61, 62, 63, 64, 65
	0, 72, 73, 74	71, 72, 73, 74, 75
	81, 82, 83, 84, 85	81, 82, 83, 84, 85

* developed as double structure

~ lost during the first year of life

analyzed in detail. Samples were taken from dentin, enamel and the border between them.

The measurements were based on two to four probes each and two different techniques were applied. No differences could be stated when compared to the same teeth of healthy children of the same age in case of the three types of samples.

Patient 2:

The phenotype of the boy showed the typical peculiarities of Down-syndrome. The chromosome analysis was performed after lymphocyte culture and confirmed a trisomy 21 (karyotype: 47,XY,+21, QFQ)

Tooth analyses:

The dentition was delayed compared to the general population. The morphology showed hypoplasia and deformation, pegtop tooth formation, and taurodontic root development. The structure of the teeth was altered. While the relation of enamel, dentin and pulpa was in the normal range, the dentin had an irregular structure (Fig. 5, Fig. 6, Fig. 7). The composition of the teeth – here the same elements were analyzed as in patient 1 and were the same in patient 1 and the control group.

DISCUSSION

Compared to craniofacial malformations and dysplasias, the abnormalities in the development and structure of teeth are often neglected in children with chromosome syndromes. Specific orthodontic investigations have demonstrated the importance of these abnormalities when characterizing chromosome aberrations by a specific pattern of morphologic changes. Some of these alterations can be delineated from abnormalities in the early embryonic development. The comparison of findings in

Figure 3. Tooth structure in patient 1. Reduced number and partially irregular arrangement of dentin channels; enlargement 2.5x.

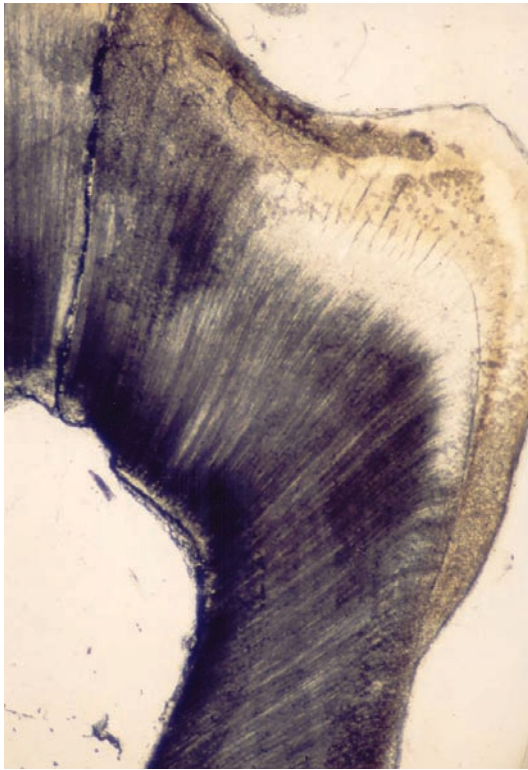


Figure 4. Simple tooth structure in patient 1 with changes in the relation of enamel, dentin and pulpa; enlargement left 1.6x, right 2.5x.

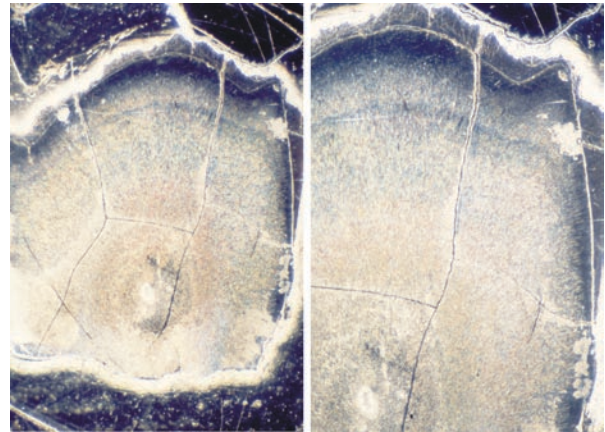


Figure 5. Patient 2 with trisomy 21 at six years of age. Irregular size and position of teeth.



children with different chromosome syndromes to the results of experimental teratogenesis in animals leads to relevant improvement of understanding the ontogenetic process [1]. Especially in patients with a cleft palate caused by different exogenous and genetic factors as well as in mice treated with different teratogenic agents leading to a cleft in the maxilla, the development of double structures of the incisives is a common symptom as in our patient with trisomy 13.

The main determination time for the development of the jaw is the third week of pregnancy. Irregularities and/or retardation of the differentiation will lead to different types of malformations. Therefore, an embryo with a trisomy 13 will – compared to one with trisomy 21 – develop the more serious pattern of malformations. A number of investigations of patients with Down-syndrome revealed peculiarities of the permanent teeth [2].

Hypodontia is common, the teeth are often smaller, and especially the third molars are frequently missing. Besides, the displacement of the maxillary canines and the disturbance of tooth order and eruptive position are described. Shapira and coworkers [3] analyzed abnormalities of the permanent teeth in 34 patients with Down-syndrome. They found an agenesis of the third molars in 74% of the patients, canine impaction in 15% and canine to first premolar transposition in 15%. All parameters were significantly higher than in the general population. In 55% of the individuals, all 4 molars were missing, followed by 35% with 2 missing molars.

There were no differences between right and left side or between the sexes. From the patients with missing molars, 60% had at least one other tooth missing and 25% had small or peg-shaped incisors, indicating that the abnormalities documented are not isolated alterations but reflect the multiple changes of the permanent teeth of patients with Down-syndrome in the same way as we observed for the first teeth.

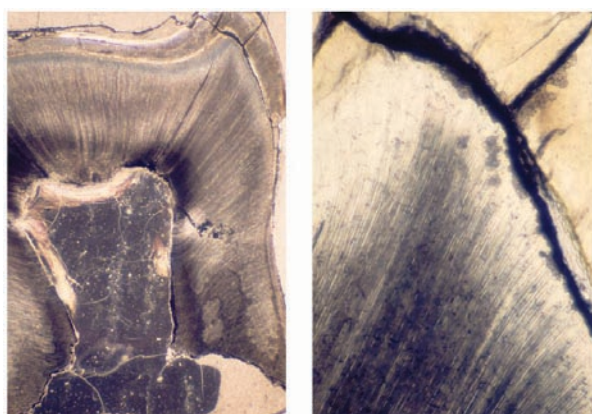
A study on the root canal anatomy of permanent teeth from patients with Down-syndrome was performed by Kelsen et al. [4]. They analyzed 281 anterior and permanent teeth of 66 individuals. They found that crown and root length, except for root length of the first premolars, was significantly shorter than in the general population. These peculiarities were also observed by other investigation groups [5]. The type of root canal was simple in the majority of teeth, canal abnormalities were not observed. The authors concluded that their findings on the size and root structure of anterior and premolar permanent teeth in patients with Down-syndrome will lead to a better orthodontic treatment in future.

In our investigation group, a further patient with trisomy 13 (karyotype: 47,XX,+13) was analyzed; but, in this case,

Figure 6. Panoramic radiograph of patient 2 at the age of six years. Taurodontia of the molars.



Figure 7. Tooth structure in patient 2. Irregular structure of the dentin channels; enlargement left 1.6x, right 2.5x.



no tooth structure investigation was possible. A follow up of dentition could only be performed until the age of 2 years and 9 month. The following peculiarities were observed: an incisor (right 51) was missing and only its bud was present. Dentition, especially of the distal teeth, was retarded.

In 1972 Gardner and coworkers [6] published the oral and dental findings in 2 cases of trisomy 13. The first one was a male fetus at term. Twenty tooth buds were removed. In the right maxilla, the lateral incisor was absent, while, on the left, a supernumerary incisor was present. The first and second molars showed the same morphology, both with missing ridges and hypoconus. The same morphologic peculiarities were observed in tooth buds from the mandibula. In the second fetus, 20 tooth buds were removed and they showed identical changes in morphology as in fetus 1.

CONCLUSION

It could be demonstrated by the present investigations and by review of findings from the literature that tooth development in children with a chromosome syndrome can deviate

significantly from the normal differentiation in the general population. Thus, there exists a new and relevant complex of symptoms to extend the characterization of rare chromosome syndromes in the future. The combination of these specific and independent peculiarities opens a new field of cooperation in clinical genetics for human geneticists, pediatricians and orthodontists.

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