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Developmental Failure of One or More Teeth in Families

Familial missing teeth

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Introduction

Failure of tooth development at the bud stage causes tooth agenesis; isolated tooth agenesis is one of the most common developmental anomalies of human permanent dentition; tooth agenesis tends to run in families, may aggregate within families, suggesting a genetic cause. Excluding the third molar, several terms are used to describe tooth agenesis: hypodontia is the absence of less than six teeth, oligodontia (severe hypodontia) is the agenesis of six or more permanent teeth and anodontia is complete absence of teeth [1, 2, 3]. Treatment depends on the severity, location, aesthetic and functional problems of missing teeth [2]. The treatment of patients affected by severe hypodontia is complex and expensive and may involve a combination of pediatric dentistry, orthodontics, prosthodontics, and implantology [4].

Objectives

Study aims: to evaluate the pattern of missing teeth in families, to observe similarities and differences of dental phenotype among affected relatives, to characterize the mode of inheritance and to identify distinct groups of patients for further molecular investigations.

Material and Methods

Clinical examinations were carried out on 26 Caucasian patients from 11 families with a family history of missing teeth. Combined examination of clinical phenotypes and panoramic radiographs improved the precision of diagnosis. Family study was used to determine whether there is a hereditary basis for tooth agenesis.

Results

In all families, the patients and their affected relatives did not share the same pattern of missing teeth. There were differences according to the tooth type, region, symmetry and number of teeth involved even in the same family. Most cases with tooth agenesis lack one or two permanent teeth.The most common missing teeth were: upper lateral incisors, second lower and upper premolars, lower central incisors, upper canines. Symmetrical hypodontia and anterior tooth agenesis were predominant. Anomalies of tooth-size and tooth-shape were observed in association with hypodontia phenotype. For further molecular investigations two groups were identified: 1. normal deciduous dentition / abnormal permanent dentition; 2. both abnormal primary and permanent dentitions.

Family 1 mother to daughter transmission	**MOTHER I2**	**DAUGHTER II1**	**DAUGTHER II2** (proband)
Age (years)	34	12	10
Hypodontia (number of missing permanent teeth)	Unilateral hypodontia (1)	Bilateral hypodontia (2)	Bilateral (severe) hypodontia (8)
Maxilla (missing teeth)	Left lateral incisor / peg-shaped right lateral incisor	Upper lateral incisors	Upper lateral incisors, canines, second premolars
Mandible (missing teeth)	–	–	Second premolars

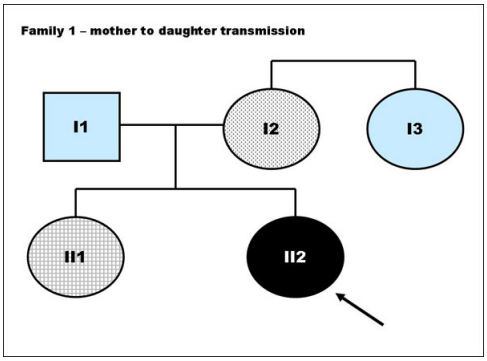


Fig 1: family 1 – pedigree

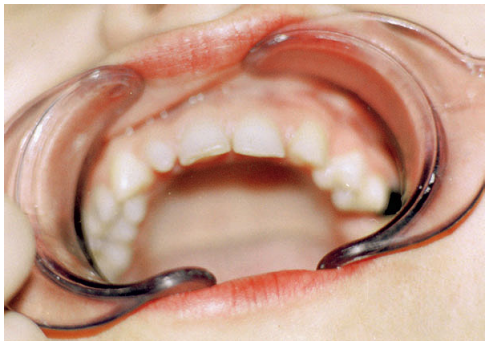


Fig 2: family 1 mother

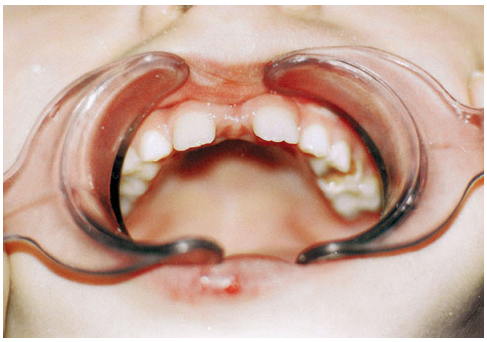


Fig 3: family 1 daughter II2

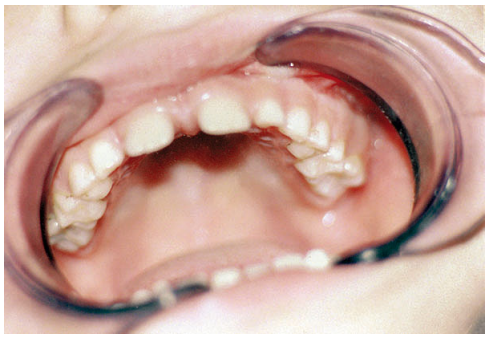


Fig 4: family 1 daughter II1

Family 2 father to son transmission	**GRANDFATHER I1**	**FATHER II1**	**SON III2** (proband)
Age (years)	79	56	27
Hypodontia (number of missing permanent teeth)	Bilateral hypodontia (2)	Bilateral hypodontia (2)	Bilateral hypodontia (2)
Mandible (missing teeth)	Central incisors	Central incisors	Central incisors

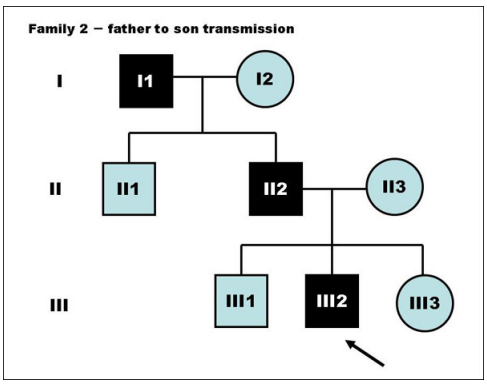


Fig 5: family 2 – pedigree

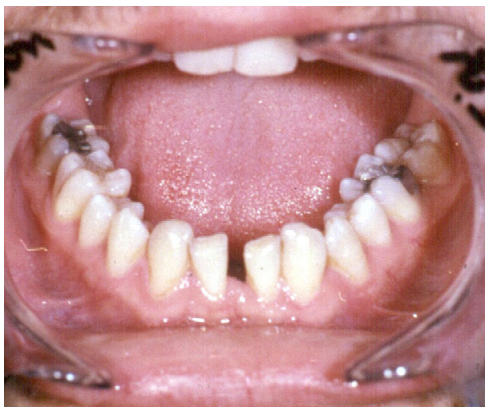


Fig 6: proband III2 intraoral view – congenitally missing lower central incisors

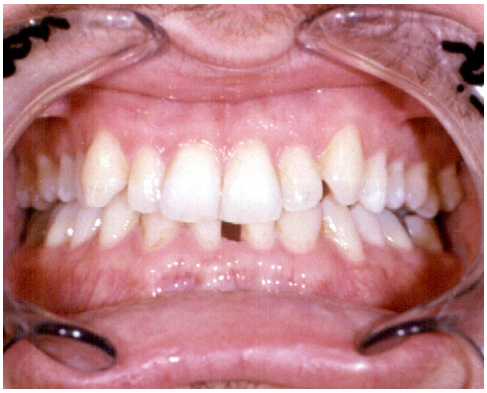


Fig 7: proband III2 oral view with tooth agenesis of lower central incisors

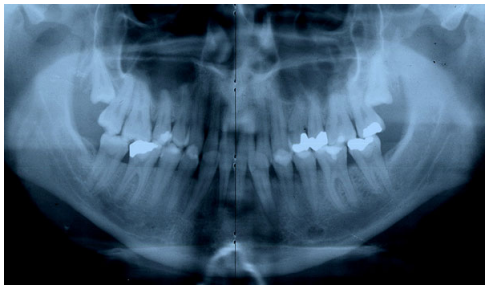


Fig 8: proband III2 – orthopantomogram

Family 3 father to daughter transmission	**FATHER I2**	**DAUGHTER II1** (proband)	**OTHER FAMILY MEMBERS (first cousin) II2**
Age (years)	36	12	12
Hypodontia (number of missing permanent teeth)	Bilateral hypodontia (2)	Unilateral hypodontia (1)	Bilateral hypodontia (2)
Maxilla (missing teeth)	–	Left lateral incisors / peg-shaped right lateral incisor	Lateral incisors
Mandible (missing teeth)	Second premolars	–	–

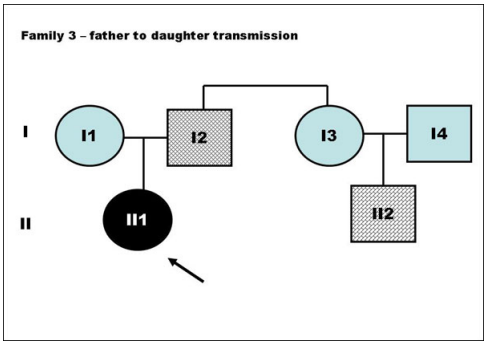


Fig 9: family 3 – pedigree

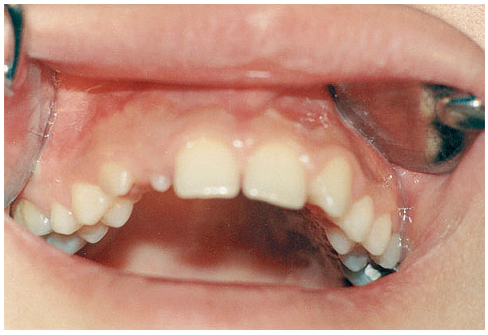


Fig 10: proband II1 – oral view: congenital agenesis of upper left lateral incisor associated with peg-shaped right lateral incicisor

Family 4 mother to daughter and son transmission	**MOTHER II2**	**DAUGHTER III2** (proband)	**SON III3**
Age (years)	36	13	10
Hypodontia (number of missing permanent teeth)	Bilateral hypodontia (6)	Bilateral hypodontia (16)	Bilateral hypodontia (6)
Maxilla (missing teeth)	Lateral incisors, second prmeolars	Central and lateral incisors, canines, first premolars	Lateral incisors
Mandible (missing teeth)	Second premolars	Central and lateral incisors, canines, first premolars	Lateral incisors, canines

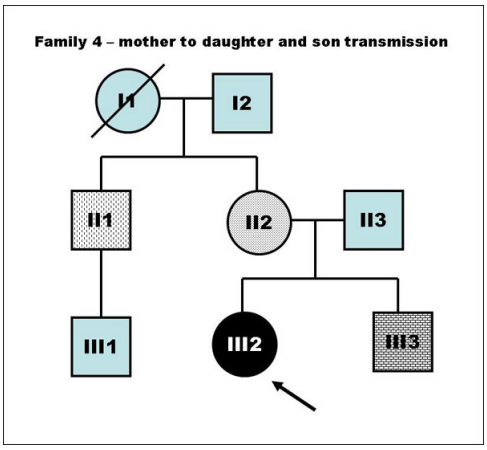


Fig 11: family 4 – pedigree

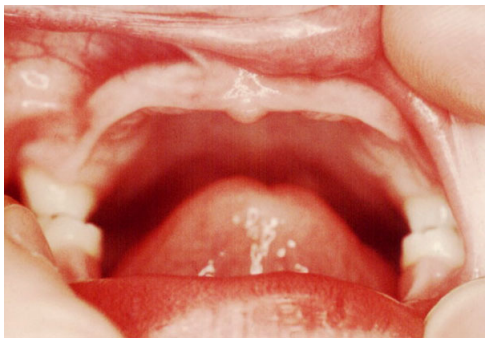
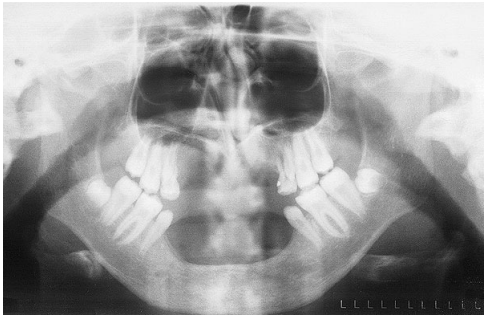


Fig 12: daughter III2 – oral view: bilateral severe hypodontia



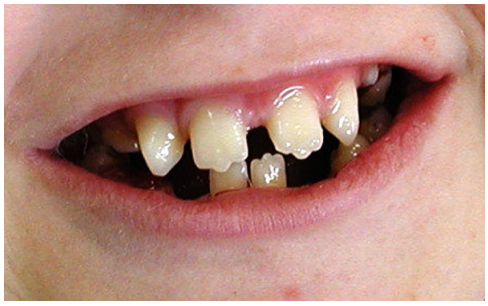


Fig 13: daughter III2 orthopantomogram

Fig 14: son III3 – oral view: bilateral hypodontia (congenital lack of upper lateral incisors, lower lateral incisors and lower canines)

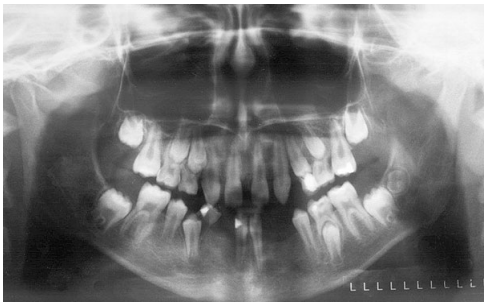


Fig 15: son III3 – orthopantomogram

Conclusions

Individuals within the same family would be expected to have the exactly mutant genes and their different dental profiles could demonstrate the gene expression variation. Peg-shaped lateral incisors may reflect incomplete expression of a gene defect that causes tooth agenesis. Unilateral tooth agenesis may be a result of reduce penetrance on the affected (right or left) side of the jaw.Familial isolated tooth agenesis is a genetic trait with clinical heterogeneity and autosomal dominant inheritance pattern.

Literature

1. Bailleul-Forestier I., Molla M., Verloes A., Berdal A.: The genetic basis of inherited anomalies of the teeth: Part I: Clinical and molecular aspects of non-syndromic dental disorders, European Journal of Medical Genetics, 2008, 51, 273-291.
2. Arte S.: Phenotypic and Genotypic features of Familial Hypodontia. Doctoral Thesis. Helsinki, Finland: University of Helsinki; 2001.
3. Stevenson R.E., Hall J.G., Human Malformations and Related Anomalies, Second Edition, Oxford university Press, 2005, 425-463.
4. Vastardis H. The genetics of human tooth agenesis: new discoveries for understanding dental anomalies. American Journal of Orthodontics and Dentofacial Orthopedics, 2000; 117: 650-656.
5. Hu JC-C., Simmer JP. : Developmental biology and genetics of dental malformations, Orthodontics&Craniofacial Research, 2007, 10 (2), 45-52.
6. Thesleff I. The genetic basis of tooth development and dental defects. Am J Med Genet A. 2006 (Dec); 140(23):2530-5.
7. Daugaard-Jensen J, Nodal M, Skovgaard LT, Kjaer I. Comparison of the pattern of agenesis in the primary and permanent dentitions in a population characterized by agenesis in the primary dentition. International Journal of Paediatric Dentistry, 1997; 7: 143-148.
8. Pemberton TJ, Das P and Patel PI, Hypodontia: Genetics and future perspectives. Brazilian Journal of Oral Science, 2005; 4:695-706.

This Poster was submitted by Prof. Dr. Emilia Severin.

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Poster Faksimile:

DEVELOPMENTAL FAILURE OF ONE OR MORE TEETH IN FAMILIES

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Background:
Failure of tooth development at the bud stage causes tooth agenesis; isolated tooth agenesis is one of the most common developmental anomalies of human permanent dentition; tooth agenesis tends to run in families (may aggregate within families) suggesting a genetic cause.

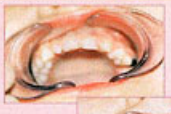
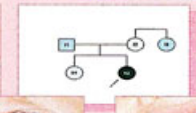
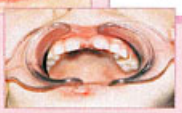
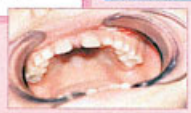
Objectives:
Our study aims are to evaluate the pattern of missing teeth in families, to examine the correlations (similarities and differences of dental phenotype) among family members to assess familial clustering, to characterize the mode of inheritance and to identify distinct groups of patients for further molecular investigations.

Patients and Methods:
Clinical examinations were carried out on 26 Caucasian patients (11 males and 15 females aged between 7 and 36) from 11 families with a family history of missing teeth. Combined examination of clinical phenotypes and panoramic radiographs improved the precision of diagnosis. Family study was used to determine whether there is a hereditary basis for tooth agenesis. We selected for the poster presentation only four families using correlation of dental phenotype between mother-daughter, father-daughter, brother-sister and sister-sister relationships.

Results
In all families, the patients and their affected relatives did not share the same pattern of missing teeth. There were differences according to the tooth type, region, symmetry and number of teeth involved even in the same family. The most commonly missing teeth were: upper lateral incisors, second lower premolars, lower central incisors, upper canines. Symmetrical hypodontia and anterior tooth agenesis were predominant. Anomalies of tooth-size and tooth-shape were observed in association with hypodontia phenotype. Except two cases, phenotypic severity did not increased in successive generation. No significant gender difference was found. Pedigree analysis indicated that congenital absence of teeth is a single-gene defect, transmitted as an autosomal dominant trait with complete penetrance and variable expressivity.

Cases of familial tooth agenesis

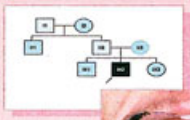
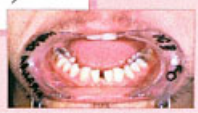
Family 1 –mother to daughter transmission	MOTHER I2 (age: 34 years)	Daughter II1 (age:12 years)	Daughter II2 (proband) (age: 10 years)
Hypodontia (number of missing permanent teeth)	Unilateral hypodontia (1)	Bilateral hypodontia (2)	Bilateral (severe) hypodontia (8)
Maxilla (missing teeth)	Left lateral incisor / peg-shaped right lateral incisor	Upper lateral incisors	Upper lateral incisors, canines, second premolars
Mandible (missing teeth)	-	-	Second premolars

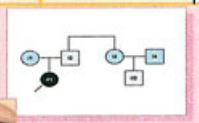
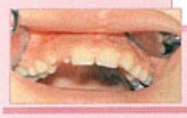
Mode of inheritance: autosomal dominant with complete penetrance and variable expression.

Family 2 – father to son transmission	Grandfather I1	Father II2	Son (grandson) III2 proband
Age (year)	79	56	27
Hypodontia (number of missing permanent teeth)	Bilateral hypodontia (2)	Bilateral hypodontia (2)	Bilateral hypodontia (2)
Mandible (missing teeth)	Central incisors	Central incisors	Central incisors

Family 3	Father I2 (age: 36 years)	Daughter II1 (proband) (age: 12 years)	Other family members (first cousin II3) (age: 12 years)
Hypodontia (number of missing permanent teeth)	Bilateral hypodontia (2)	Unilateral hypodontia (1)	Bilateral hypodontia (2)
Maxilla (missing teeth)	-	Left lateral incisor / peg-shaped right lateral incisor	Lateral incisors
Mandible (missing teeth)	Second premolars	-	-

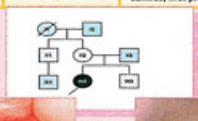



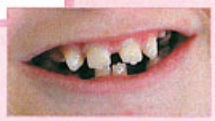
Mode of inheritance: autosomal dominant with complete penetrance and constant expressivity.

Mode of inheritance: autosomal dominant with complete penetrance and variable expressivity.

Family 4: mother to daughter and son transmission	Mother II2 (age: 36 years)	Daughter III2 (age: 13 yrs)	Son III3 (age: 10 years)
Hypodontia (number of missing permanent teeth)	Bilateral hypodontia (6)	Bilateral hypodontia (16)	Bilateral hypodontia (6)
Maxilla (missing teeth)	Lateral incisors, second premolars	Central and lateral incisors, canines, first premolars	Lateral incisors
Mandible (missing teeth)	Second premolars	Central and lateral incisors, canines, first premolars	Lateral incisors, canines





Mode of inheritance: autosomal dominant with complete penetrance and variable expressivity.

Discussion and Conclussions
Individuals within the same family would be expected to have the exact mutant genes and their different phenotypes could demonstrate the gene expression variation. Peg lateral incisors may reflect incomplete expression of a gene defect that causes tooth agenesis. Unilateral agenesis may be a result of reduced penetrance on the side affected.
Familial isolated tooth agenesis is a genetic trait with clinical heterogeneity and autosomal dominant inheritance pattern.