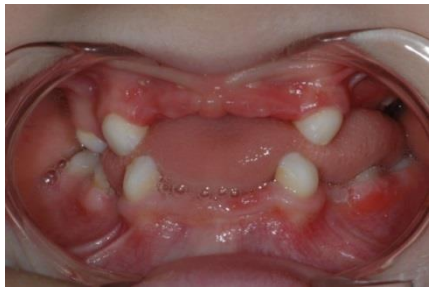
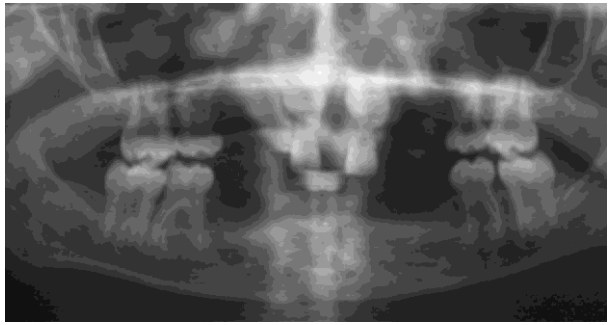
the Child Dental Patient with

OLIGODONTIA

clinical, genetic and therapeutic aspects

F Clauss, M Strub, M-C Manière





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Faculté de chirurgie dentaire Université de Strasbourg

Les Hôpitaux Universitaires de STRASBOURG

European Reference Networks

Oligodontia : definition and prevalence

- Dental anomaly defined by at least **6 dental ageneses in primary or permanent dentitions**
- Genetic etiology with marked intra-familial phenotypic variability
- Epidemiology: low prevalency between 0.08% and 0.16%
- Oligodontia are highly **polygenic** : numerous genes involved in molecular etiology
- Mendelian inheritance is autosomal dominant in most of the cases
- Isolated forms with only dental phenotypic manifestations
- Associated to minor dermatological signs : *WNT10A* gene mutations

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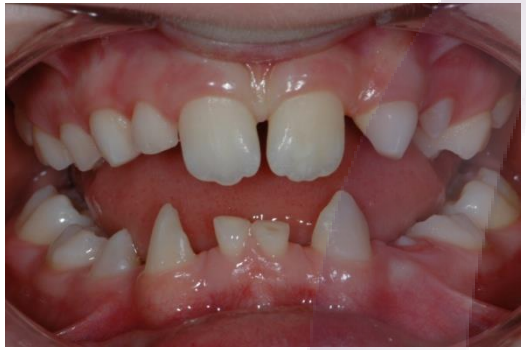
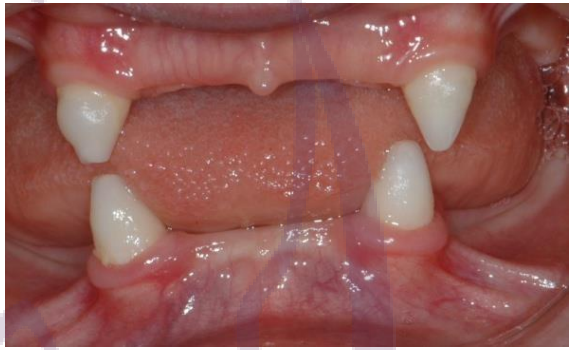
Isolated and syndromic oligodontia

Dental phenotype defined by a minimum of 6 ageneses



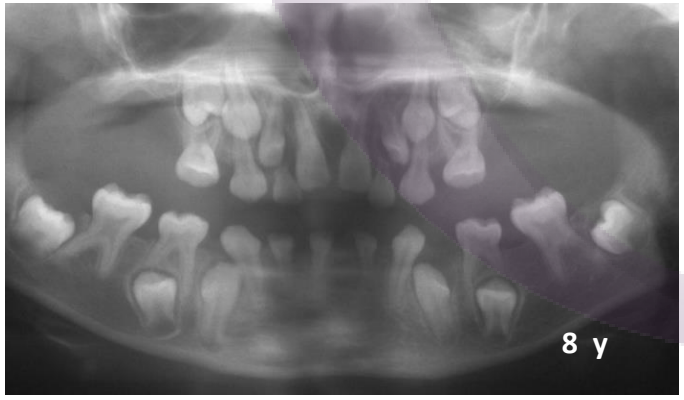


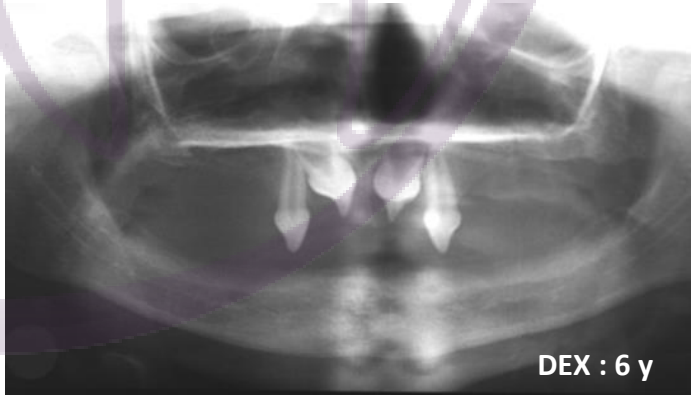
Isolated oligodontia: mutation in

- *MSX1*
- *PAX9*
- *AXIN2*
- *EDA-EDARADD-WNT10A*

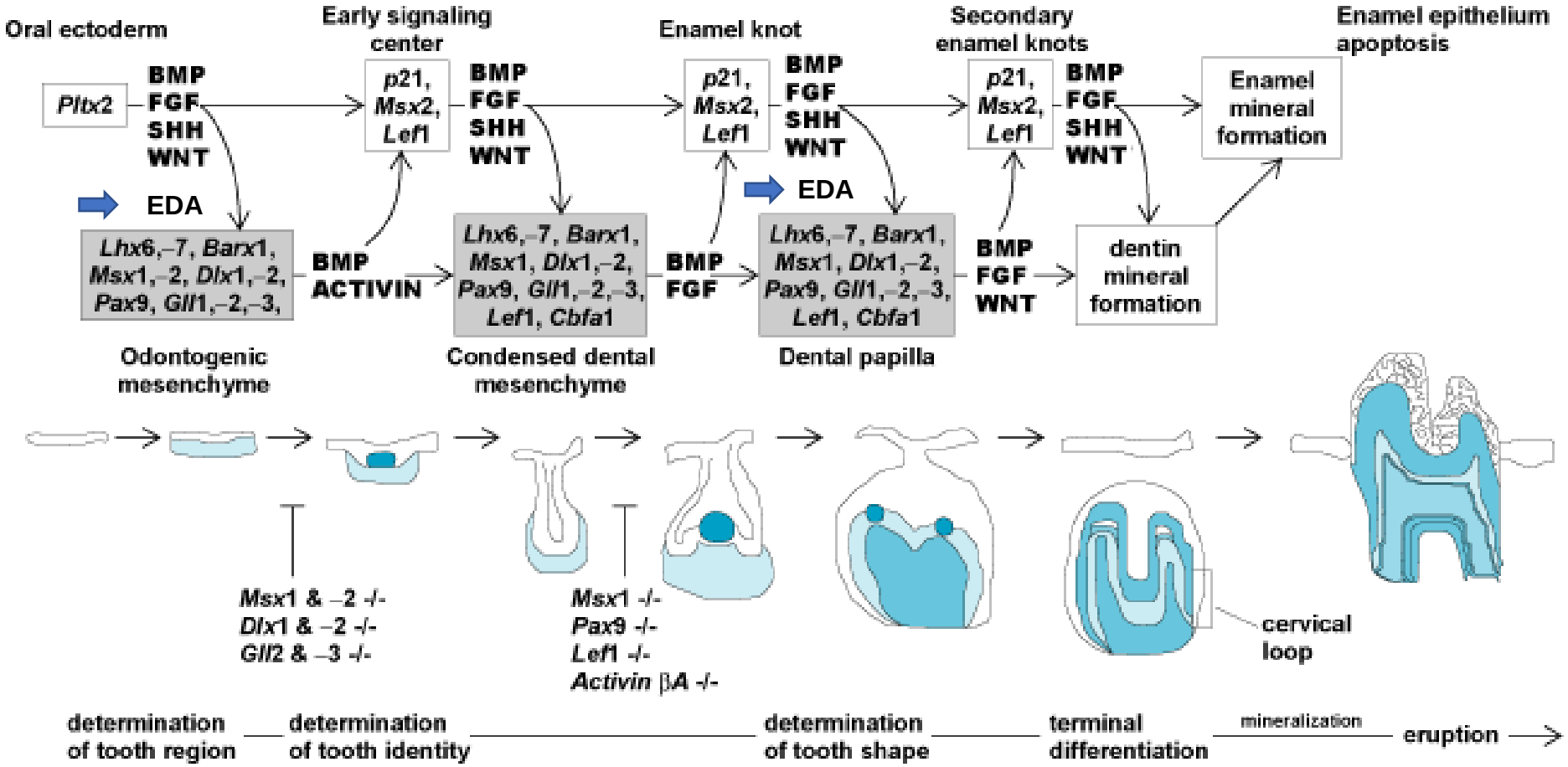
Syndromic oligodontia

- Hypohidrotic Ectodermal Dysplasia (HED)
- Rieger, Wolff-Hirschorn, Van der Woude, Johanson-Blizzard syndromes, Craniosynostoses…





Odontogenesis and molecular etiologies of oligodontia



Thesleff, 2006

Oligodontia : isolated forms

- Dental ageneses with specific topographic distribution depending of the gene mutated : **phenotype-genotype correlations**
- Ageneses associated to absence or delay in dental eruption : clinical sign for early detection in primary dentition
- Ageneses of incisors or premolars : potential association with *MSX1* or *WNT10A* mutations
- Ageneses of molars : potential association with *PAX9* mutations
- Frequent association between dental ageneses-morphological anomalies-malpositions

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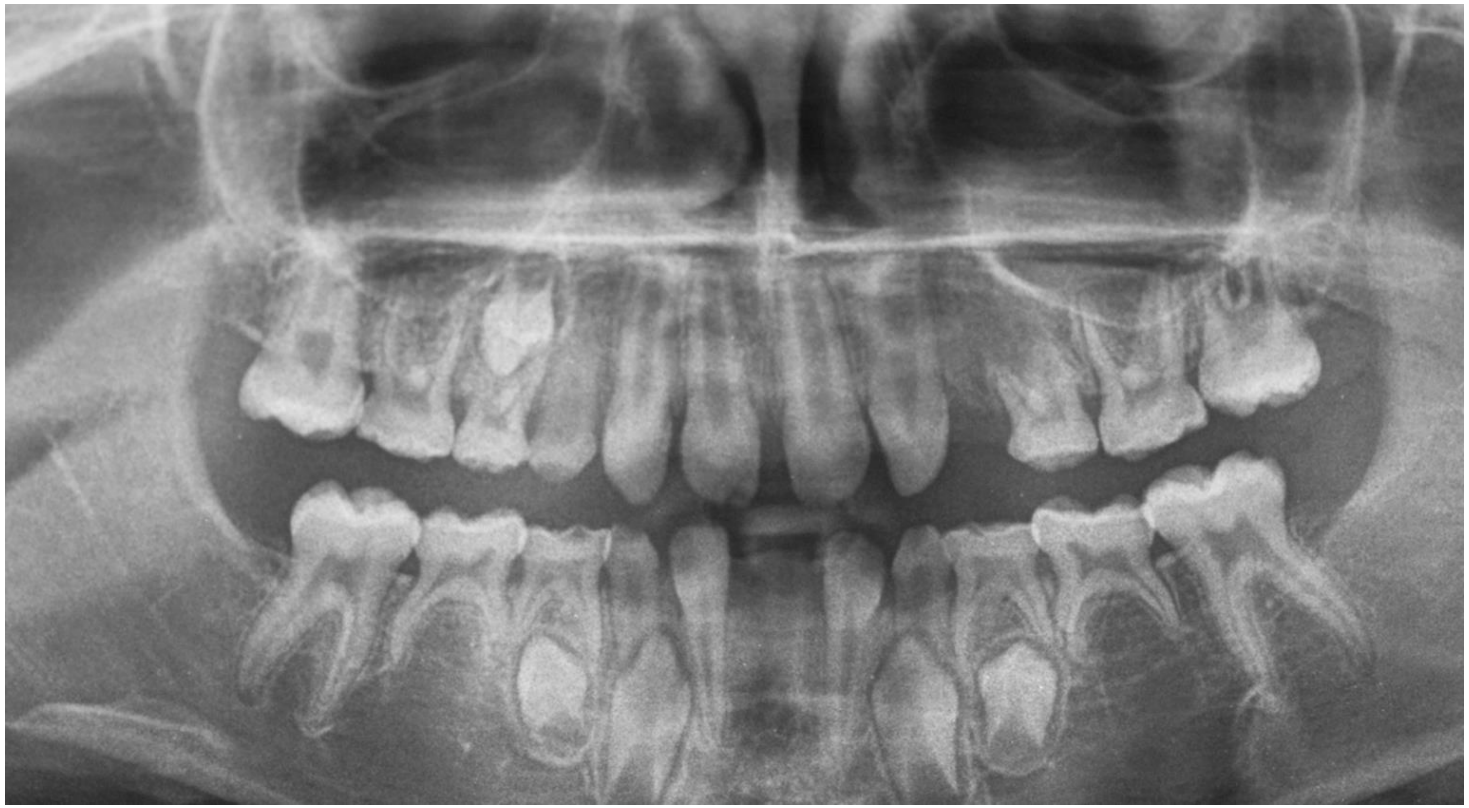
Oligodontia : radiological features of isolated forms



8 year-old patient
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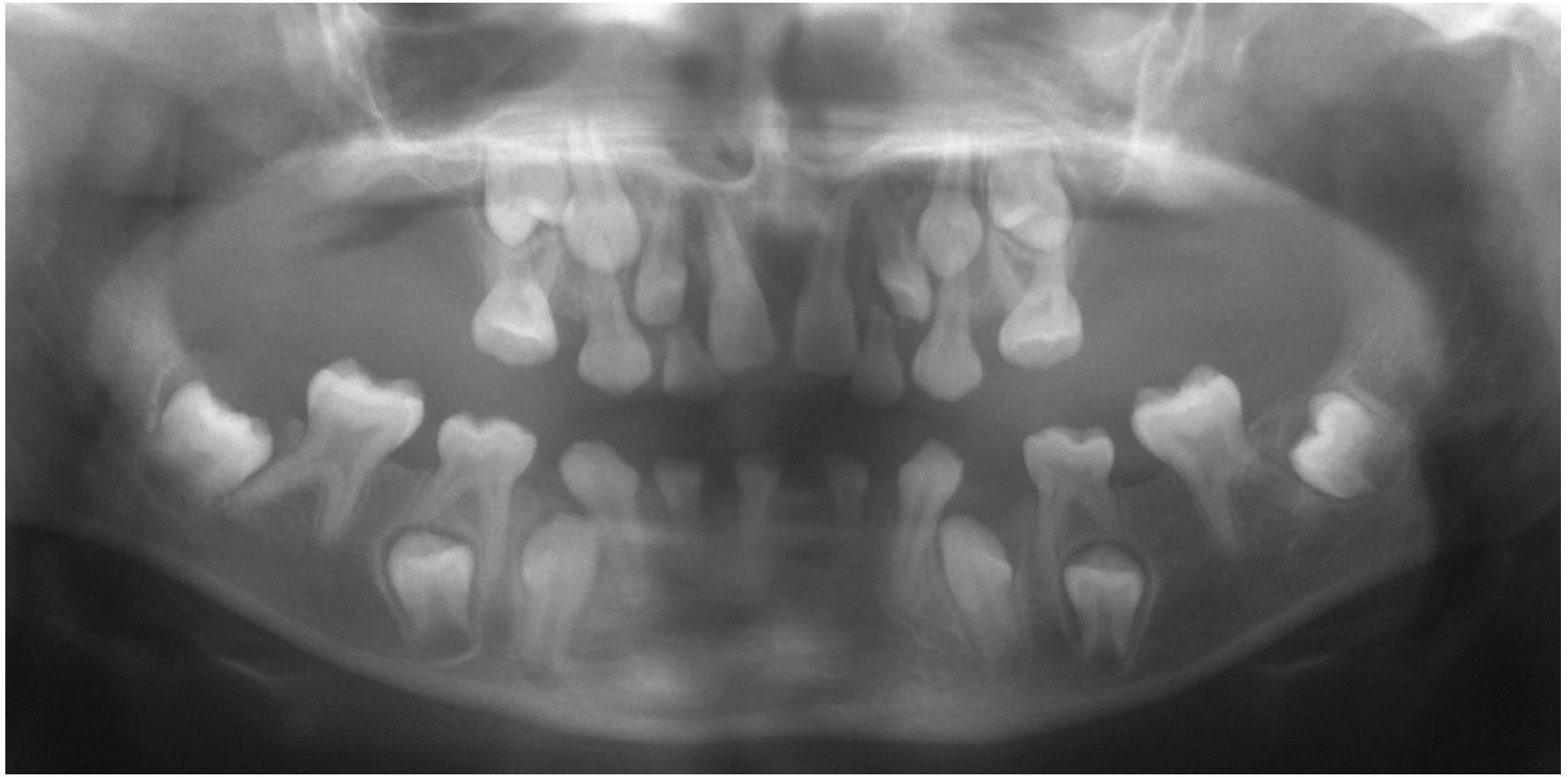
Ageneses of lateral maxillary incisors, premolars, mandibular incisors and molars

Oligodontia : radiological features of isolated forms



Severe phenotype with ageneses of mandibular incisors, premolars and molars :
potential *PAX9* mutation

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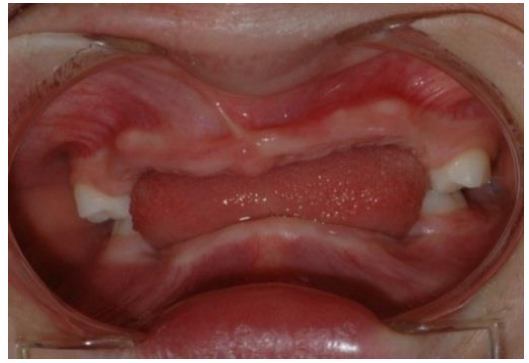
Oligodontia : syndromic forms

- **Syndromic oligodontia** : in association with other systemic manifestations (neurological, skeletal, ophtalmological, cardiac defects) : *P63* syndromes, Rieger syndrome (*PITX2* mutation), Wolf-Hirschorn syndrome (*MSX1* deletion)

- Most common forms of syndromic oligodontia : **ectodermal dysplasia (ED)** characterized by the clinical associations **oligodontia-hypotrichosis-hypohidrosis** (sweat glands ageneses)

- Most prevalent : X-linked Hypohidrotic Ectodermal dysplasia

- Autosomal dominant and recessive forms of Ectodermal Dysplasia

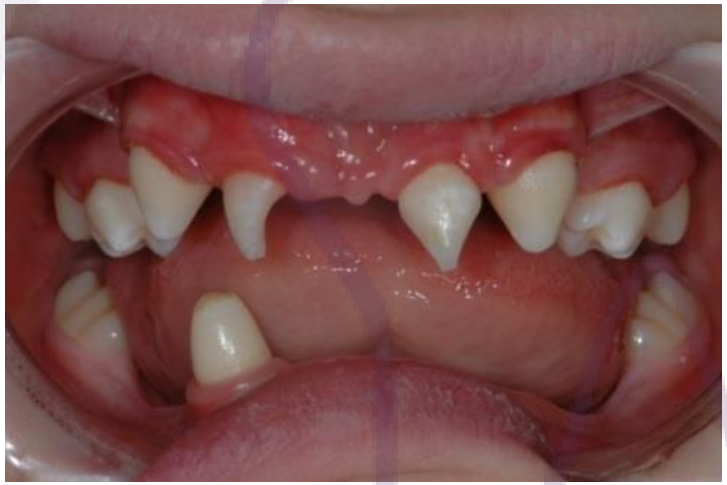


Syndromic oligodontia : group of ectodermal dysplasia

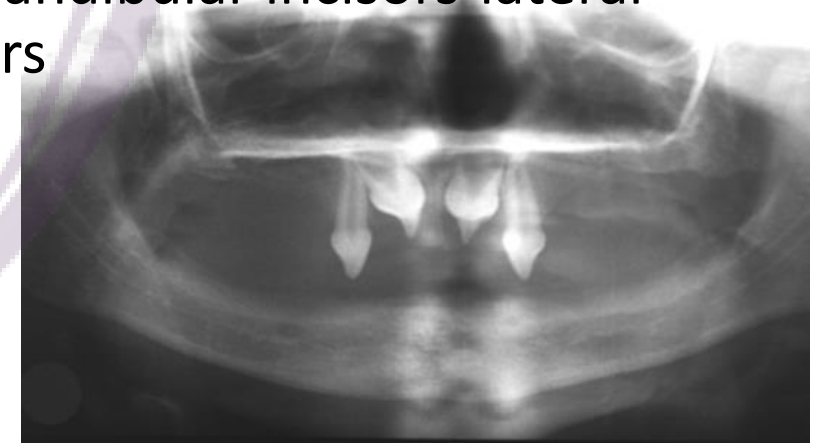
- Distinction beween ectodermal dysplasia with only ectodermal features (X-linked and autosomal ED or with extra-ectodermal defects)
- ED with neurological or immune defects : *Incontinentia Pigmenti* – ED with immunodeficiency and osteopetrosis
- ED with skeletal manifestations : *P63* syndromes, tricho-dento-osseous syndrome
- ED with endocrine pathologies
- ED associated to deafness or cleft lip-palate

Syndromic oligodontia : group of ectodermal dysplasia

- Dental phenotype associated to X-linked ED : ageneses-cone-shaped teeth (incisors and canines)

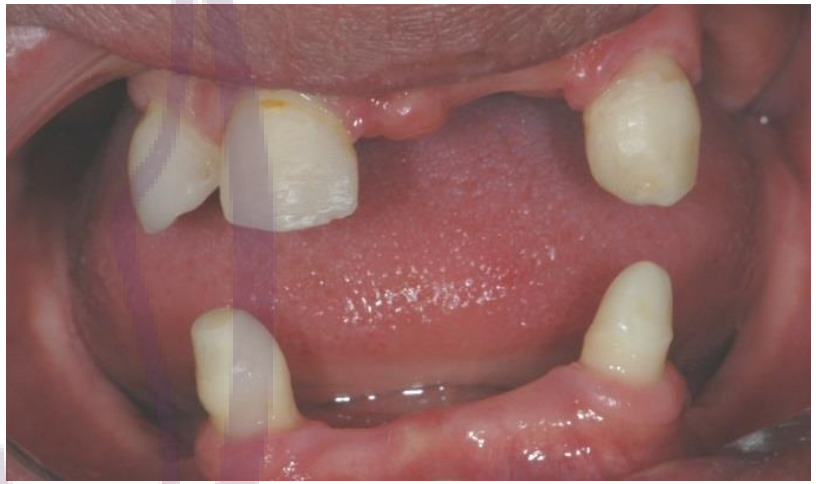


- **Most frequent dental ageneses in primary dentition** : mandibular incisors-lateral maxillary incisors-mandibular molars-first maxillary molars
- Average of 8 dental ageneses in primary dentition



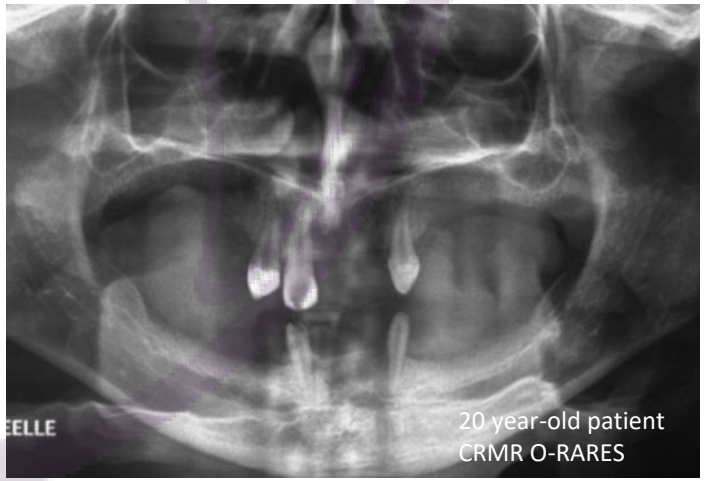
Syndromic oligodontia : group of ectodermal dysplasia

- **Most frequent dental ageneses in permanent dentition in X-linked ED** : mandibular incisors-lateral maxillary incisors-second mandibular premolars-second maxillary molars-second molars



- Average of dental agenesis: between 11 and 16 missing teeth

- Severe phenotype of oligodontia associated to anomalies of eruption and taurodontism



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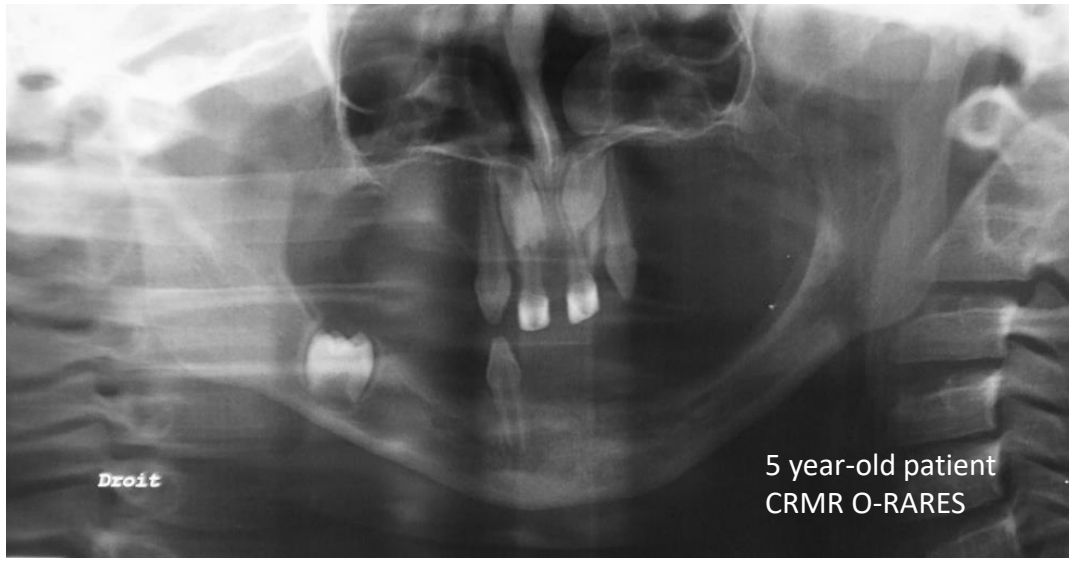
Radiological features of X-linked ED



5 year-old patient
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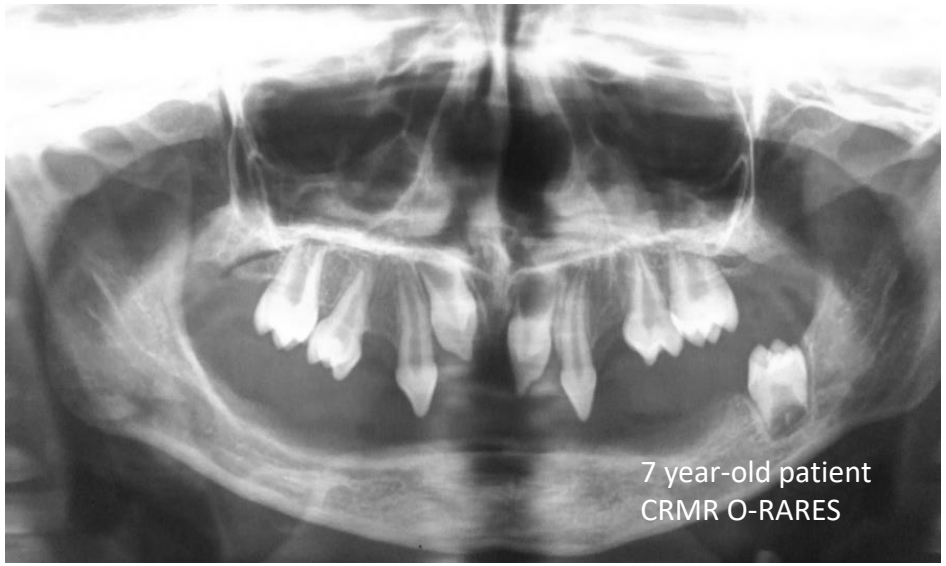
7 year-old patient
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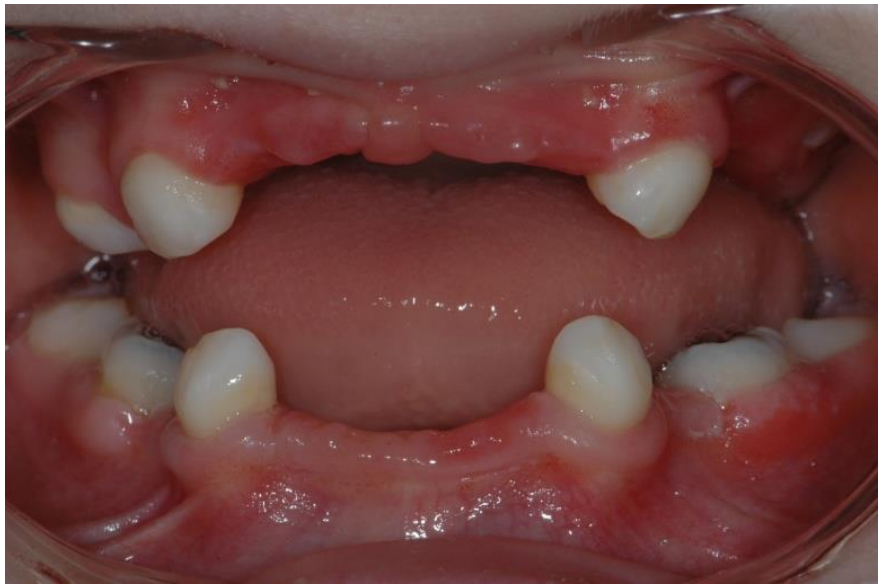
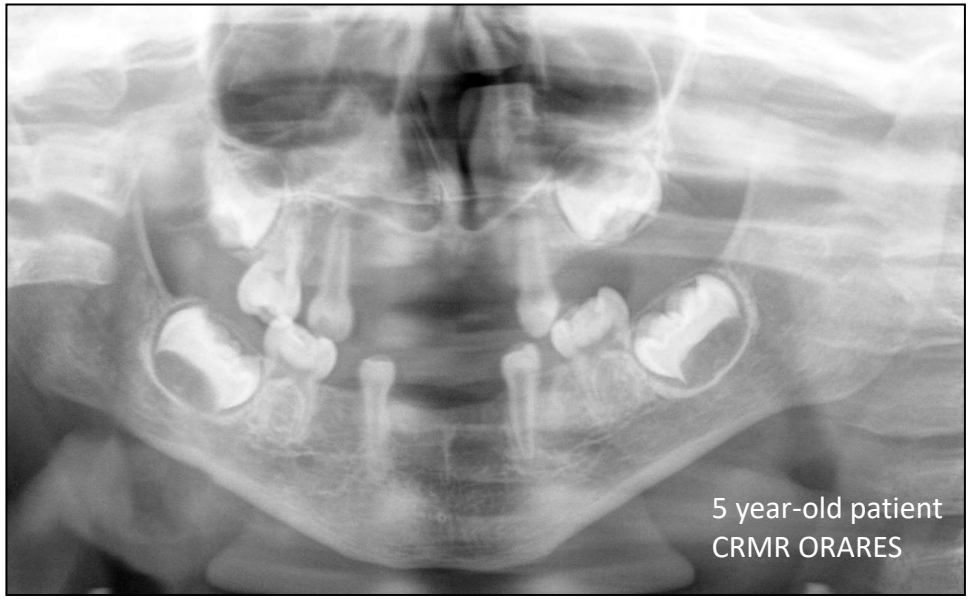
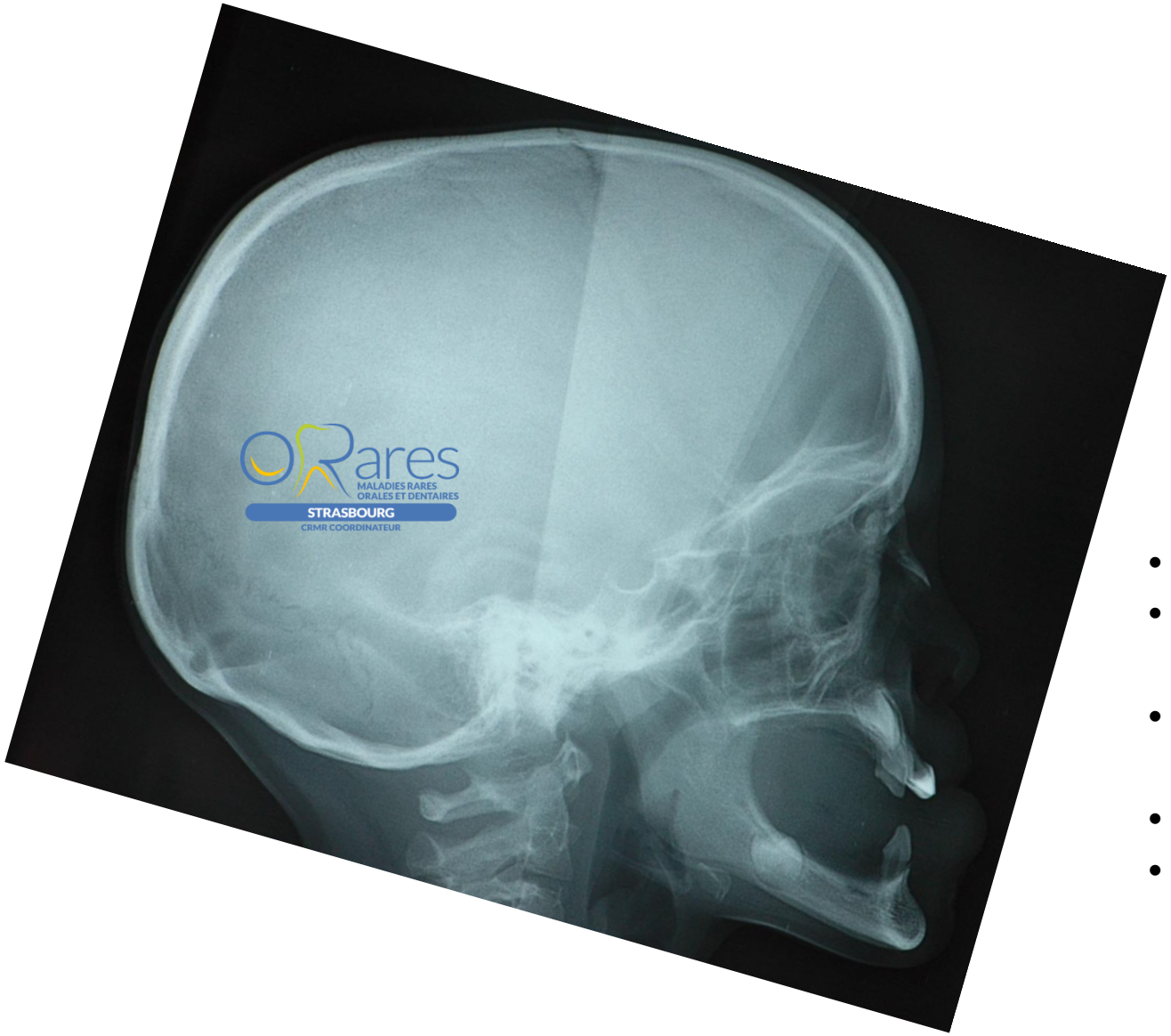
Clinical and radiological features of autosomal dominant ED





Cranio-facial manifestations associated to X-linked ED : radiological features observed on profile teleradiography



- Maxillary hypoplasia
- Mandibular prognathism
- Hypotrophic alveolar bone
- Frontal prominence
- Cranial base defects

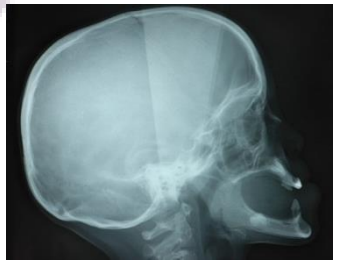
Dental,medical and molecular diagnosis in the context of ED

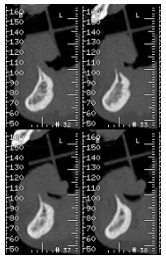
- **Meticulous medical and familial anamnesis** : presence of other systemic manifestations, persons affected in the family, dental phenotype observed in parents, brothers or sisters of the patient

- **Extra-oral examination**: dysmorphosis associated to ED (facial concavity, maxillary hypoplasia, skeletal class III)

- **Dental examination**: number and topographic distribution of dental ageneses (determination of the phenotypic severity), morphological anomalies, degree of hypotrophy of the mandibular alveolar crest

- **Complementary exams:** panoramic radiograph, teleradiography, cone-beam computed tomography (early implants treatment),
  hand and wrist x-ray (skeletal maturity)





Dental, medical and molecular diagnosis in the context of ED

- **Consultation in a medical genetic department**: molecular and biological explorations (analysis of *EDA-EDAR-EDARADD-WNT10A* mutations), type of intra-familial transmission

- **Genetic counselling**: determination of the risks of transmission

- **Multi-disciplinary diagnosis and management**: ENT, pneumology, ophtalmology, gastro-enterology..

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Early prosthetic and implant treatments in ED

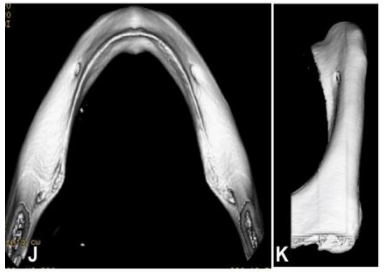
- **Conventional prosthetic treatment** : from the age of 3-4 years old depending of the maturity and cooperation

- Regular prosthetic follow-up with adaptation to the skeletal growth and dental eruption

- Severe phenotypes of mandibular oligodontia with previous prosthetic failure : indication of early implants therapy

Early prosthetic and implant treatments in ED

- **Early implants treatment in ED**: placement from the age of 6 years old of two symphyseal mandibular implants
- **Importance of pre-operative CBCT** for determination of mandibular bone morphology and dimensions
- Placement of implants in the symphyseal area under general anesthesia for young patients
- Stabilization of an implants-supported removable mandibular prosthesis
- Follow-up till the end of the growth and implants placement in the maxilla and posterior mandibular sectors



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