CONGRESUL CONSACRAT ANIVERSĂRII A 75-A DE LA FONDAREA USMF „NICOLAE TESTEMIȚANU”
20 23 octombrie 2020

# CLINICAL AND GENETIC PARTICULARITIES IN FRANCESCHETTI SYNDROME: PRESENTATION OF CLINICAL CASE

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## Introduction

Franceschetti syndrome (FS) also known as Treacher-Collins syndrome (TCS), is a congenital disorder of craniofacial development characterized by bilateral symmetrical oto-mandibular dysplasia, without abnormalities of the extremities and associated with several head and neck defects. It affects both genders equally. FS can be inherited in an autosomal dominant or autosomal recessive manner.

Autosomal dominant-about 55%-61% of probands have the disorder as the result of a de novo TCOF1, POLR1D, or POLR1B pathogenic variant. At conception, each sib of an affected individual has a 25% chance of being affected, a 50% chance of being an asymptomatic carrier, and a 25% chance of being unaffected and not a carrier.

**Keywords:** *Franceschetti syndrome, child, hypoplasia, mandible*

## Purpose

The importance of clinical and genetic approaches to Franceschetti syndrome is to establish an early diagnosis and present the clinical case.

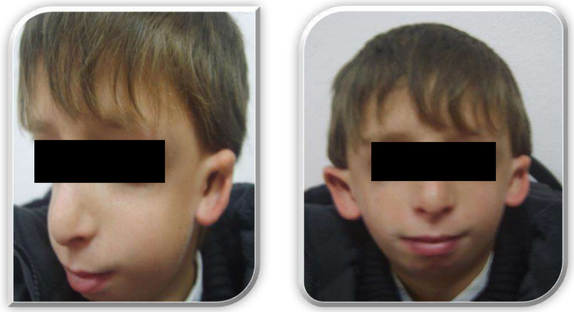
**Differential diagnosis**

| Disorder | Gene | MOI | Distinguishing Clinical Features |
|---|---|---|---|
| **Mandibulofacial dysostosis with microcephaly** | EFTUD2 | AD | •Microcephaly<br>•ID<br>•Asymmetry of facial features<br>•Esophageal atresia<br>•Thumb abnormalities |
| **Nager syndrome** | SF3B4 | AD | Limb deformities, preaxial abnormalities (e.g., small or absent thumbs, triphalangeal thumbs, radial hypoplasia or aplasia, radioulnar synostosis) |
| **Miller syndrome** | DHODH | AR | Limb deformities, postaxial abnormalities (e.g., small or absent 5th digit incl 5th metacarpal, ulnar hypoplasia, absent 5th toe) |
| **Toriello-Carey syndrome** | | AR | •Failure to grow<br>•Microcephaly<br>•Agenesis of corpus callosu<br>•Urogenital anomalies in affected males<br>•Facial dysmorphisms (hypertelorism, flattened nasal bridge, anteverted nares)<br>•Short neck |

## Material and methods:

this paper comprises analysis of the bibliographic sources regarding children that have Franceschetti syndrome and the synthesis of the clinical case of a 9-year-old boy with FS.

**Case 1. Franceschetti syndrome**
A 9-year-old boy



## Results:

A 9-year-old boy applied for a clinical-genetic evaluation during the medical-genetic consultation at the Mother and Child Institute. Clinical examination: underdevelopment of facial bones and mandibular hypoplasia, characteristic appearance of the face - facies in "bird's beak", macrostomy, the inclination extends below the external angle of the eyelid slit - antimongoloid appearance, nazofrontal angle is obliterated and the nasal bone is increased, bilateral malformations of the ears, hearing loss up to 40%, normal intelligence. The diagnosis was established based on clinical criteria, based on the association of characteristic dysmorphism and transmission deafness. Radiological examination revealed typical changes of the mandible. The audiogram confirmed bilateral transmission deafness.

## Conclusions

1. The knowledge of the clinical-genetic aspects according to the literature contributes to the early diagnosis of FS in children.
2. Identification of the mutation in the FS is at the research stage, but molecular study by the indirect method can contribute to genetic counseling in familial forms.
3. Treatment should be tailored to the specific needs of each individual, preferably by a multidisciplinary craniofacial management team.

**Selective Bibliography:**

1.https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3214538/
2.https://ghr.nlm.nih.gov/condition/treacher-collins-syndrome
3.https://www.ncbi.nlm.nih.gov/books/NBK153/