

Bilateral Digital Clubbing in a 10 years Old Boy

Jorge Sales Marques^{1,2*}

¹*Pediatric Department, Centro Hospitalar Conde S. Januário, Macau, China*

²*Pediatric Department, Centro Hospitalar Vila Nova De Gaia/Espinho, Portugal*

***Corresponding Author:** Jorge Sales Marques, Pediatric Department, Centro Hospitalar Conde S. Januário, Macau, China.

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Abstract

Clubbing often occurs in heart and lung diseases and is caused by congenital heart defect, chronic lung diseases like bronchiectasis, cystic fibrosis or lung abscess.

Congenital digital clubbing (CDC) is a genetic disorder with recessive inheritance and the gene identified is HPGD (hydroxy prostaglandin dehydrogenase 15-NAD).

The clinical features are consisted with bilateral hands and feet digital clubbing, without any pain, with normal heart and lungs screening.

We reported a 10-year-old boy with bilateral digital clubbing since two years ago.

The subsidiary examination showed normal blood test for liver, thyroid and celiac diseases, with chest x - ray and heart ultrasound without any changes.

The HPGD gene was negative for CDC, but with his clinical presentation and negative results for diseases than can cause pathologic digital clubbing, we can classify this case as CDC.

No treatment is needed in CDC patients.

Genetic counselling is offered for the couple because CDC is inherited as autosomal recessive disorder.

Keywords: *Bilateral Digital Clubbing; Congenital*

Background

- When we find a patient with digital clubbing, the first diagnosis in our mind is a case secondary to heart or lungs diseases.
- But this clinical presentation is not always caused by these situations.
- Congenital digital clubbing (CDC) is a genetic disorder with recessive inheritance, first described in 1919 in two unrelated families [1].
- The chromosome affected is 4q34.1 and the gene identified is HPGD (hydroxy prostaglandin dehydrogenase 15-NAD) [2].

- The clinical characteristic is consisted with bilateral hand and feet digital clubbing, without any pain, with normal heart and lungs screening.

Case Report

- A 10 years old boy was observed in the consultation because of hand and feet digital clubbing, with signs beginning two years before (Figure 1).
- No history of pain, exercise intolerance, cough, respiratory distress, dyspnea or orthopnea, easy tiredness, asthenia or difficulty in climbing stairs.
- The parents denied any family history of same changes. They both have normal fingers.
- On physical examination, the child showed normal phenotype, normal pulmonary auscultation, no heart murmur and hepatosplenomegaly.
- The hand and feet showed bilateral digital clubbing, without any pain on touching.
- No other change was noted.
- The clinical findings were consisting of congenital digital clubbing.
- Blood test revealed normal for liver enzymes, thyroid function and transglutaminase and gliadin antibodies.
- The heart ultrasound and chest x- ray were also normal.
- The study of the HPGD gene was negative in our case.



Figure 1: Bilateral digital clubbing of the hands.

Discussion

- Clubbing often occurs in heart and lung diseases and is caused by congenital heart defect, chronic lung diseases like bronchiectasis, cystic fibrosis or lung abscess.
- Others non heart or lung problems can also affect the digital fingers: celiac disease, cirrhosis, Graves’ disease and Hodgkin lymphoma.
- CDC results from proliferation of the connective tissues between the nail matrix and the distal phalanx and abnormal function of the nail matrix.
- One Japanese study has also suggested a possible role of auto-immune factors in the causation of clubbing shows the pathogenesis of clubbing (Figure 2) [3].
- Clinically, sometimes the feet toes are spared but the fingers are always involved.
- The prevalence is unknown.
- The confirmation of the disease is by the molecular study of HPGD gene.
- When the gene result is negative, we cannot exclude the disorder.
- With the screening of heart, lungs, liver, thyroid and celiac diseases normal, we can classify this as CDC, even with the HPGD gene negative.
- No treatment is needed in this case.
- Genetic counselling is offered for the couple because CDC is inherited as autosomal recessive disorder.

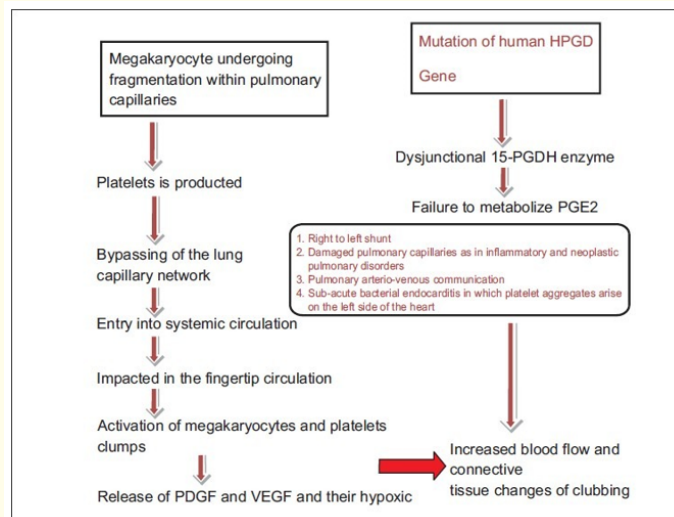


Figure 2: Pathogenesis of digital clubbing.

Conclusion

Digital clubbing not always is secondary to heart or lung diseases, because can be congenital related to HPGD gene.

Bibliography

1. Weber FP. "The occurrence of clubbed fingers in healthy persons as a familial peculiarity". *British Medical Journal* 2 (1919): 379.
2. Tariq M., *et al.* "Mutation in the HPGD gene encoding NAD⁺ dependent 15-hydroxyprostaglandin dehydrogenase underlies isolated congenital nail clubbing (ICNC)". *Medical genetics* 46 (2009): 14-20.
3. DM Mahesh. "Digital clubbing Malay Sarkar". *Lung India* 29.4 (2012): 354-362.

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