

# MOTHER AND BABY DIAGNOSED NOONAN SYNDROME WITH DYSMORPHIC FINDINGS

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## OBJECTIVE

Noonan syndrome is an autosomal dominant genetic disorder characterized by short stature, low hair line, webbed neck, cubitus valgus, chest wall deformities and congenital heart defects. Here; the patient was admitted to hospital by parents due to undescended testis and finally infant and mother were diagnosed Noonan syndrome.

## CASE

A 14-month-old male patient was admitted to hospital because of bilateral undescended testis. On physical examination there was growth failure. Body weight 7.5 kg (<3p), height 69 cm (<3p). There were also dysmorphic face, hypertelorism, ptosis, bilateral undescended testis. Cytogenetic chromosome analysis of the case was 46 XY. Ultrasonography and echocardiography were normal. Genetic mutation was reported as Noonan syndrome. The genetic test also sent from the mother on observing similar dysmorphic findings. The diagnosis was again Noonan's syndrome. Orchiopexy-applied case was followed because of growth retardation.



## CONCLUSION

In this study; we showed that dysmorphic findings and undescended testis may be stimulant for the diagnosis of Noonan syndrome in the early period. The mother of the case also was diagnosed as Noonan syndrome interestingly by showing the dominant transition effect.

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