Two Different Endocrine Cancer, One Disease; DICER-1 Mutation

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INTRODUCTION

Autosomal dominant DICER1 mutations are among the causes of early-onset familial cancer. DICER-1 mutation has been shown in pleuropulmonary blastomas as well as **ovarian tumors**, **thyroid**, parathyroid, pituitary, **adrenocortical** and testicular tumors. It is important to be aware of the risk for the development of other cancers in the follow-up of these cases.
DICER-1 mutation was detected in two cases who presented with different rare endocrine tumors.

CASE-1

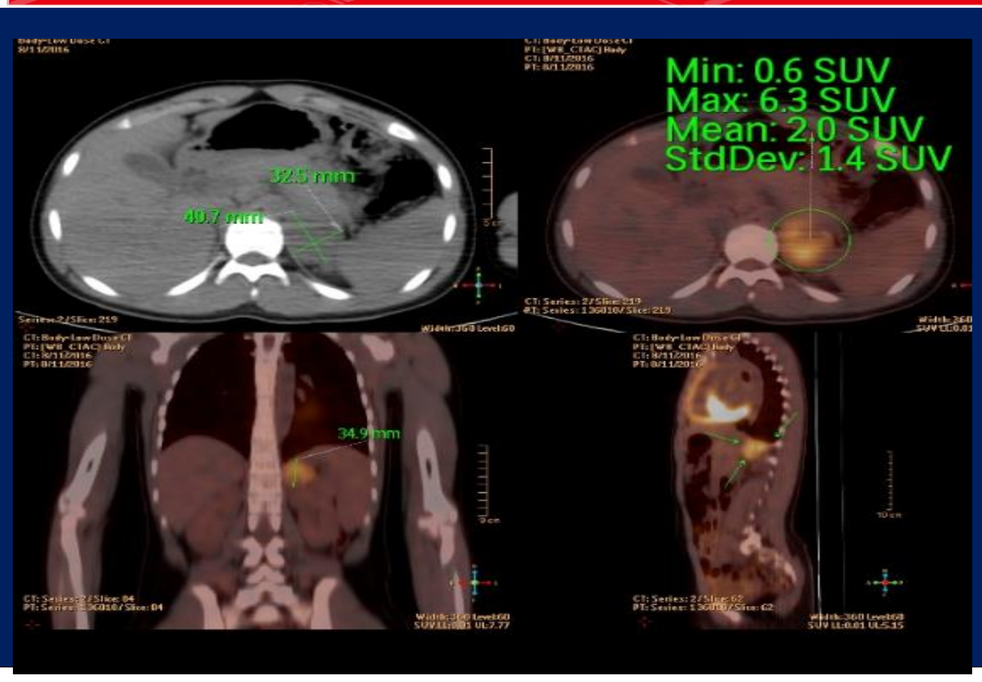
8.5 year old girl
Complaint: deepening voice and hirsutism
Family History: Parents are first degree cousins
- Aunt had a history of thyroid cancer

Physical Examination:
- Height was significantly above the familial target height percentile
- Thelarche:Tanner stage 1, pubic hair development: stage 5
- Size of the clitoris: 3x1cm
- Increased muscle mass

Laboratory evaluation: Total testosterone: 231,6 ng/dl (N<10)
USG: A mass of 43 mm in the left adrenal area
Pathologic diagnosis: Adrenocortical cancer
Treatment: Chemotherapy + Mitotane

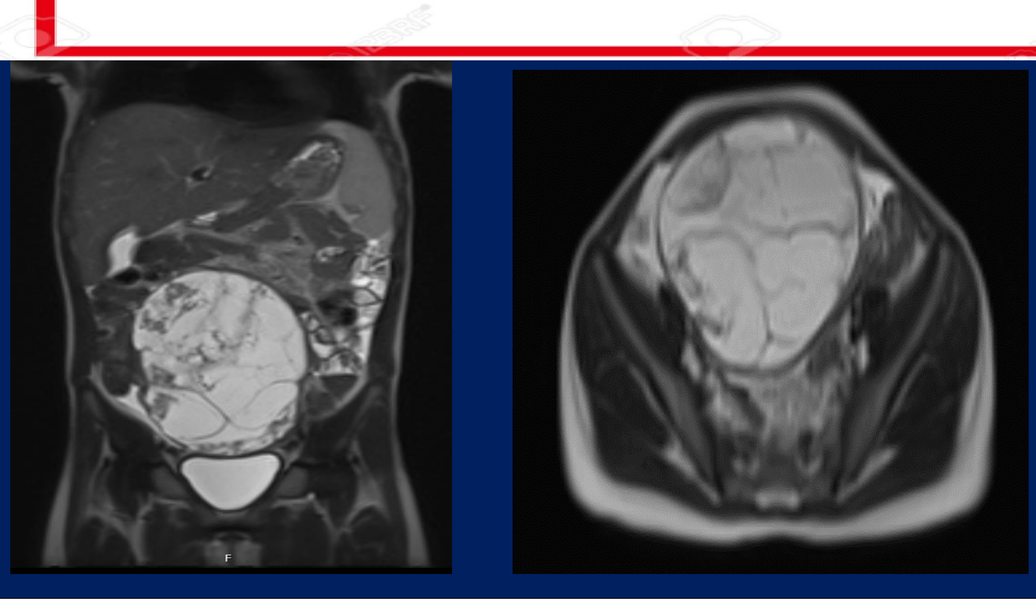
During the follow-up visits, a simple ovarian cyst with the diameter of 3cm was detected and dissappeared spontaneously

In the left surrenal area, a mass with diameters are **41x33x35 mm**
(necrotic areas are olsa present in the lesion)



	Testosterone ng/dl (N<10)	TSH mIU / L (N:0,6-4,84)	fT4 ng/dl (N:0,97-1,67)	Calsitonin pg/ml (N:0-11,5)	Thyroid USG	LDH U/L (N:135-214)	AFP IU/ml (N:0-5,8)	B-hCG Ng/ml (N:0-2)	CA-125 U/ml (N:0-35)
CASE-1	**231,6**	2,26	1,48	-	Normal	**273**/179	0,945	<2	19,87
CASE-2	<2,5	2,65	1,59	<2	8mm nodule	**681**	2,21	<2	**70,45**

A mass of **7x11x15 cm** in diameters, originate from right adnexial area
(smooth-thin contoured mass lesion with diffuse septation)



CASE-2

6.5 year old girl
Complaint: Abdominal pain
Family History: parents are second degree cousins
Physical Examination:
- A mass was palpated in the abdomen
- Pubertal development was Tanner stage 1

Laboratory evaluation:
- Elevated CA-125 and LDH levels

USG: a mass of 15 cm in diameter with right adnexial origin
Pathologic diagnosis: Sertoli-Leyding cell tumor
Treatment: Chemotherapy

During follow-up, a solid nodule (8 mm in long axis) in the left lobe of the thyroid gland was detected. Fine needle aspiration biopsy revealed a benign lesion.

CONCLUSION

As seen in our cases, DICER-1 mutation should be considered in the presence of multiple organ involvement in endocrine cancers and other endocrine organ pathologies should be kept in mind during the follow-up period.