

# Pituitary Imaging Abnormalities and Related Endocrine Disorders in Erdheim–Chester Disease

Skand Shekhar, Jorge A. Irizarry-Caro, Ninet Sinaii, William A. Gahl, Juvianee I. Estrada-Veras, Rahul H. Dave, Bernadette R. Gochuico, Georgios Z. Papadakis, Nicholas Patronas, Constantine A. Stratakis, Kevin O'Brien and Fady Hannah-Shmouni

**Supplement 1 (Table).** Hormone deficits, treatment and pituitary imaging characteristics of ECD cohort.

ECD Serial No/ Sex	Age at NIH Visit	Age at Diagnosis of ECD	Endocrine Abnormalities	Hypothalamic-Pituitary Lesion on MRI	Treatment for ECD
1-M	46	43	Central hypogonadism, DI	Small pituitary; narrowing of the pituitary stalk; loss of pituitary bright spot	Imatinib
2-M	42	42	Central AI, central hypogonadism, DI	Stalk deviation	PEG-IFN, steroids
3-F	32	32	None	Thickened pituitary stalk	PEG-IFN
4-M	64	54	Central hypogonadism	Abnormal enhancement; pituitary encasement; possible cyst intermediate; absent posterior lobe	Anakinra
5-M	47	43	Central hypogonadism	Partial empty sella	Interferon alfa
6-M	48	44	Central AI, central hypogonadism, central hypothyroidism, DI	Abnormal enhancement; absent posterior lobe; suprasellar mass causing lateral displacement of pituitary segments; loss of pituitary bright spot	Methotrexate, Remicade, Ionizing radiation
7-M	68	67	Central hypogonadism	Small pituitary; thickened pituitary stalk; tapering of the infundibulum; partially empty sella; loss of pituitary bright spot	Interferon alfa
8-M	42	36	Central hypogonadism, central hypothyroidism, DI	Enlarged pituitary stalk at hypothalamus; secondary empty sella with small pituitary; loss of pituitary bright spot	Anakinra, Interferon alfa
9-M	50	46	Central hypogonadism, DI	Thickened stalk at the base and right side of pituitary, invading the cavernous sinus; abnormal pituitary enhancement	Interferon alfa
10-M	64	63	None	None	None
11-F	46	44	DI	None	Cladribine
12-M	41	36	Central hypogonadism, DI	None	Dabrafenib, Trametinib, Interferon alfa
13-M	51	48	Central AI, central hypogonadism, central hypothyroidism, DI	Thickened stalk; pituitary encasement	Dabrafenib, Trametinib
14-M	64	57	DI	Loss of pituitary bright spot	Interferon alfa
15-M	51	46	None	Abnormal pituitary morphology; thickened pituitary stalk	Cladribine
16-M	57	58	Central AI	None	Vemurafenib
17-M	61	59	None	None	Imatinib, Dabrafenib, Trametinib
18-F	32	28	DI	Loss of pituitary bright spot	Ionizing radiation, Cladribine, Interferon alfa
19-M	57	57	None	Loss of pituitary bright spot	None
20-F	19	17	None	Abnormal pituitary morphology; thickened pituitary stalk	PEG-IFN

21-F	64	64	None	Small pituitary	Cladribine
22-F	62	56	Central hypothyroidism	Small pituitary; partially empty sella	Dabrafenib, Trametinib
23-F	50	50	Central AI, central hypogonadism, hyperprolactinemia	Cyst intermediate	6-Mercaptopurine, Vincristine
24-M	67	61	None	Small pituitary; partially empty sella	Anakinra
25-F	57	53	None	Cyst intermediate	Cladribine
26-M	39	38	Central AI, central hypogonadism, DI, hyperprolactinemia	Small pituitary; thickened pituitary stalk; loss of pituitary bright spot	Interferon alfa
27-M	34	33	Central AI, central hypogonadism, central hypothyroidism, DI	Abnormal pituitary morphology-heterogenous enhancement; small pituitary; thickened pituitary stalk; stalk deviation; microadenoma; loss of pituitary bright spot	None
28-M	51	51	Central hypogonadism, hyperprolactinemia	None	Interferon alfa
29-M	45	42	None	None	None
30-M	50	49	None	None	PEG-IFN
31-F	48	47	Central hypogonadism, DI, hyperprolactinemia	Cyst intermediate; suprasellar mass; loss of pituitary bright spot	Interferon alfa
35-M	45	44	Central hypogonadism, DI	Heterogenous enhancement of pituitary gland; small pituitary; thickened pituitary stalk; partially empty sella; loss of pituitary bright spot	Vemurafenib
36-M	61	61	Central hypogonadism	None	6-Mercaptopurine, Steroids, Vinblastine
37-M	72	69	DI	None	Anakinra
38-M	54	52	Central hypogonadism, DI	None	Interferon alfa, Dabrafenib, Trametinib
39-M	56	51	None	None	Vemurafenib
40-M	59	58	Central hypogonadism	None	Vemurafenib
41-M	37	37	None	None	Anakinra
42-M	58	57	Central hypogonadism, DI	None	Interferon alfa
43-M	55	51	Central AI, central hypogonadism, DI	Loss of pituitary bright spot	Vemurafenib
44-M	68	65	None	None	Cyclophosphamide, Steroids
45-F	30	28	Central AI, central hypogonadism, central hypothyroidism, DI, hyperprolactinemia	Thickened pituitary stalk; pituitary gland enlarged abutting the optic chiasm; loss of pituitary bright spot	Cladribine
46-M	47	39	Central AI	Pituitary was symmetrical	Remicade
47-M	44	34	DI	Loss of pituitary bright spot	Interferon alfa, dabrafenib, trametinib
48-M	61	61	Central AI, central hypogonadism	Abnormal configuration of the pituitary; thickened pituitary stalk; stalk deviation; pituitary encasement; partially empty sella; loss of pituitary bright spot	Steroids
49-M	56	56	Central AI, central hypogonadism	None	None

<b>50-M</b>	45	42	Central hypogonadism, DI	Thickened pituitary stalk	Dasatinib
<b>51-F</b>	64	63	None	Thickened pituitary stalk; stalk deviation; loss of pituitary bright spot	Anakinra
<b>52-F</b>	48	48	Central hypogonadism, hyperprolactinemia	None	PEG-IFN
<b>53-M</b>	65	65	Central hypogonadism, hyperprolactinemia	Rathke's cyst	None
<b>54-M</b>	64	61	Central hypogonadism, DI	Abnormal pituitary morphology-heterogenous enhancement; loss of pituitary bright spot	Vemurafenib
<b>55-M</b>	58	58	Central hypogonadism, DI	Loss of pituitary bright spot	Interferon alfa
<b>56-F</b>	64	61	None	None	Anakinra, Vemurafenib
<b>57-M</b>	50	46	Central hypogonadism, DI	Thickened pituitary stalk; loss of pituitary bright spot	None
<b>58-M</b>	52	52	None	Stalk deviation	Vemurafenib
<b>59-F</b>	58	58	None	Thickened pituitary stalk; stalk deviation; loss of pituitary bright spot	None
<b>60-F</b>	57	47	Central AI, hyperprolactinemia	None	Interferon alfa
<b>64-M</b>	69	68	None	None	Methotrexate
<b>67-M</b>	47	47	None	Thickened pituitary stalk; stalk deviation; loss of pituitary bright spot	Dabrafenib, Trametinib
<b>68-M</b>	74	74	Central AI, central hypogonadism, hyperprolactinemia	Loss of pituitary bright spot	Steroids
<b>71-M</b>	46	45	Central hypogonadism	None	Dabrafenib, Trametinib

AI, adrenal insufficiency; DI, diabetes insipidus; PEG-IFN, pegylated interferon.