

*Case Presentation:*  
**An Irritable Infant  
With a Surprise Diagnosis**

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# Presentation

- 2 year old female infant
- Presented to local DGH with 2 month history of
  - vomiting
  - intermittent diarrhoea
  - lethargy and loss of appetite



# Past Medical History

- Normal term delivery weight 2.99 kg
  - Unrelated parents
  - two older healthy sibs
  - occasional episodes of profuse vomiting
- otherwise nothing of note



# On Examination

- Pale
- Chest X-ray: NAD
- Abdomen difficult to palpate due to tensing
- Abdominal ultrasound reported as normal apart from loops of static bowel



# Investigations

- U&e, LFTs, unremarkable
- FBC normal except for platelets, 583 (169-358)
- Ammonia 22, lactate 1.4
- CRP 20.7 (0-8)
- Urine culture: neg



# Metabolic testing

Samples sent for metabolic screening  
Due to unexplained vomiting

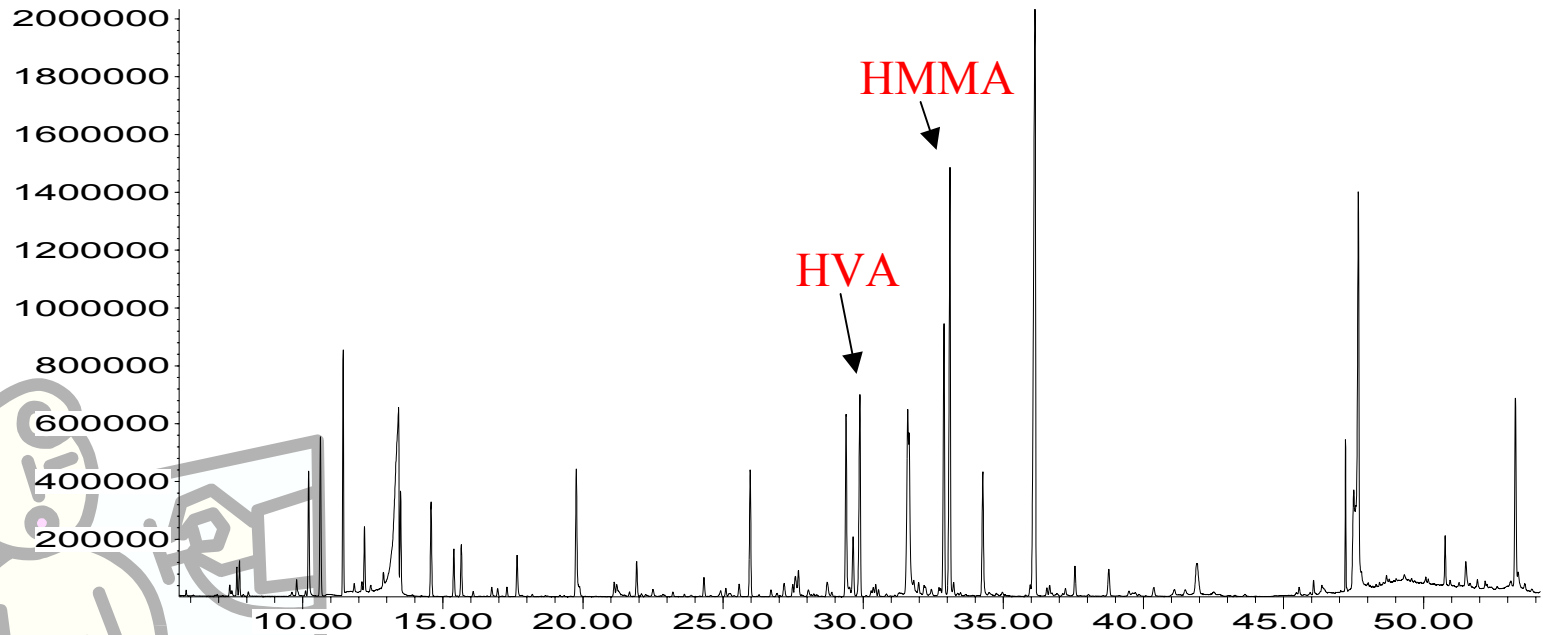
- Plasma and urine amino acids: NAD

• But.....



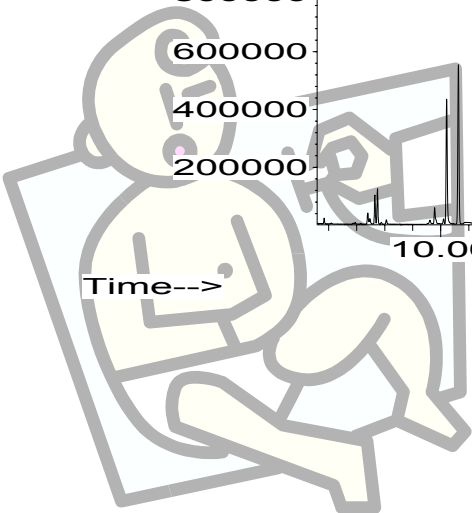
# Urine Organic Acids

Abundance



Time-->

Elevated HMMA and HVA, otherwise NAD



# Catecholamine Metabolites

Repeat samples requested for quantitation

(preserved in acid)

HVA; **86** umol/mmol creat (< 23)

HMMA; **147** umol/mmol creat (< 14)

Dopamine; **27.1** umol/mmol creat (< 1.75)

*Confirmation of elevated catecholamine excretion.*

*Not consistent with an IEM, but pathognomonic of*

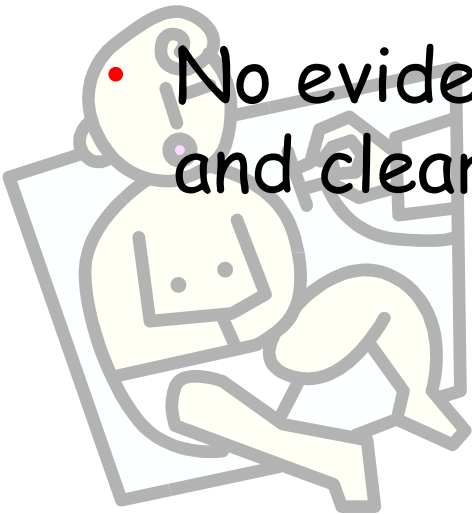
*a tumour, neuroblastoma*





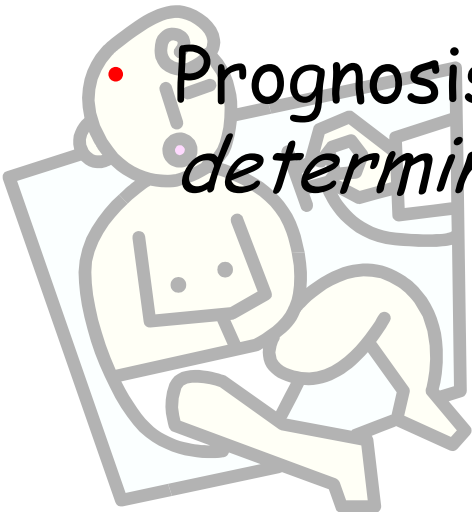
# Clinical Examination at Regional Centre

- High resolution CT showed abdominal showed calcified mass on the midline, approx 9x9 cm encasing aorta and vena cava
- Smaller sub-clavicular mass
- No evidence of metastases, bone and lungs and clear by MIBG scan



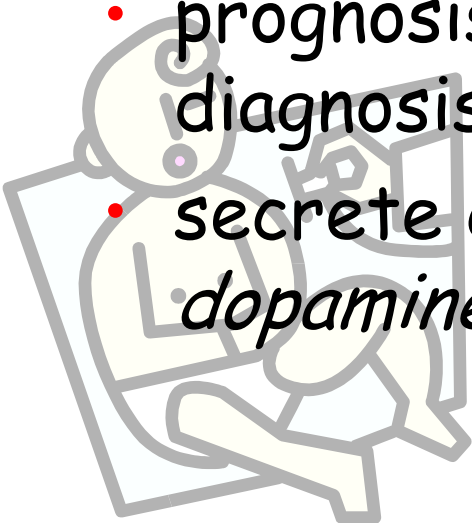
# Treatment and Prognosis

- Parents understandably distressed at diagnosis but early detection should improve prognosis
- Chemotherapy started within two weeks of initial evidence of diagnosis
- Prognosis depend on n-myc status *not yet determined*

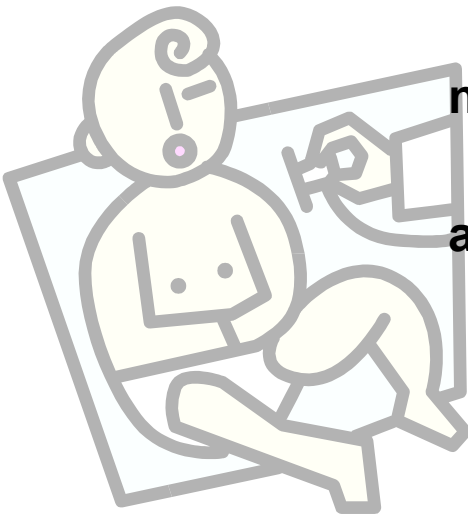
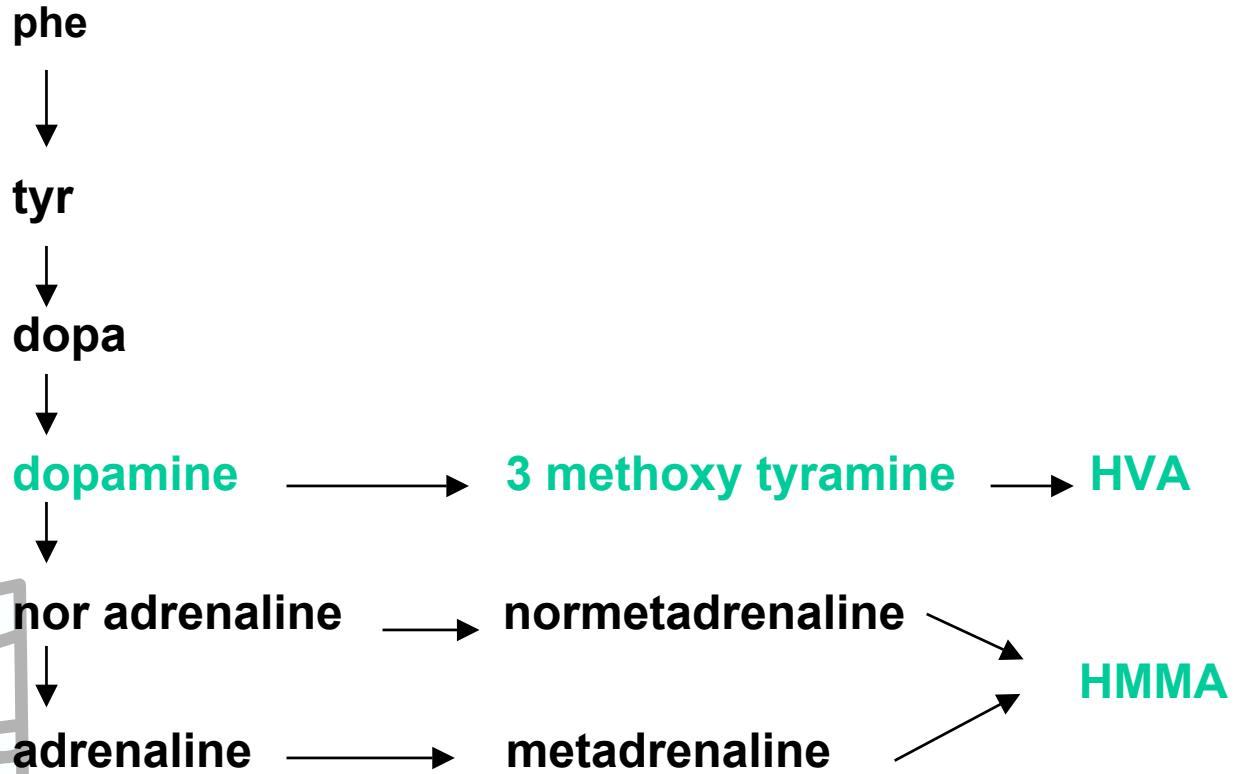


# Neuroblastoma

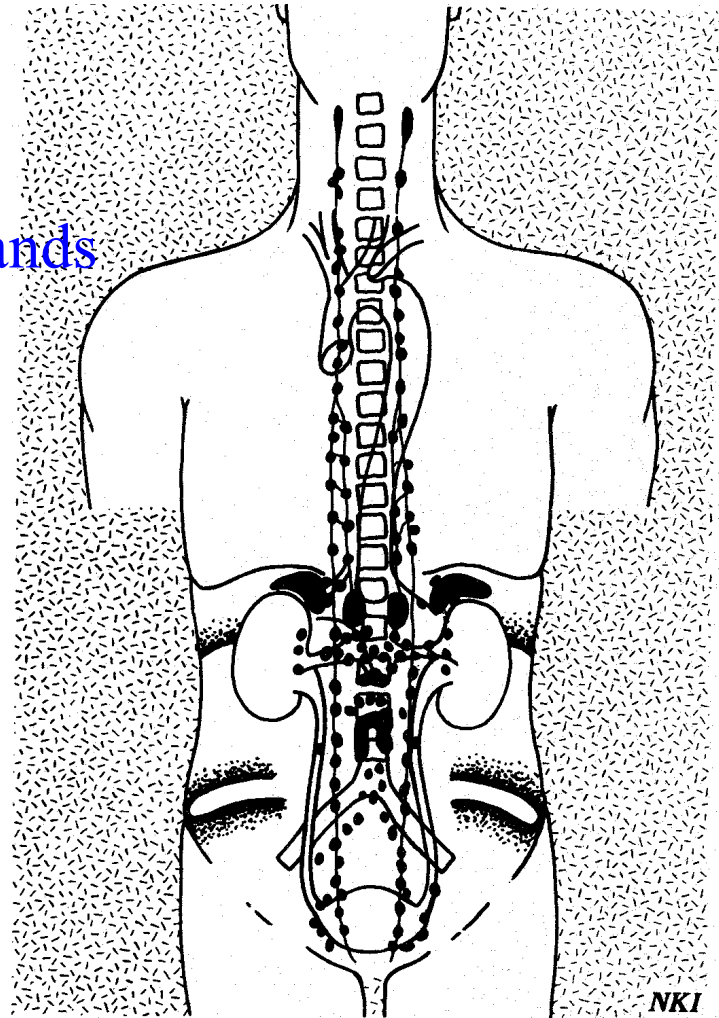
- arise from developing sympathetic nerve cells, neuroblasts
- common solid childhood tumour
- highly malignant
- prognosis relates to age and stage at diagnosis
- secrete catecholamines, *predominantly dopamine and its metabolites*



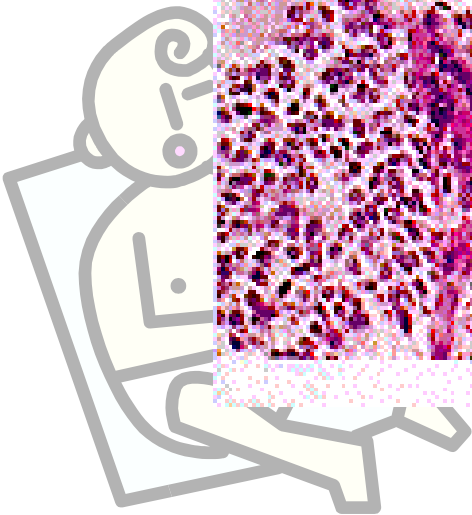
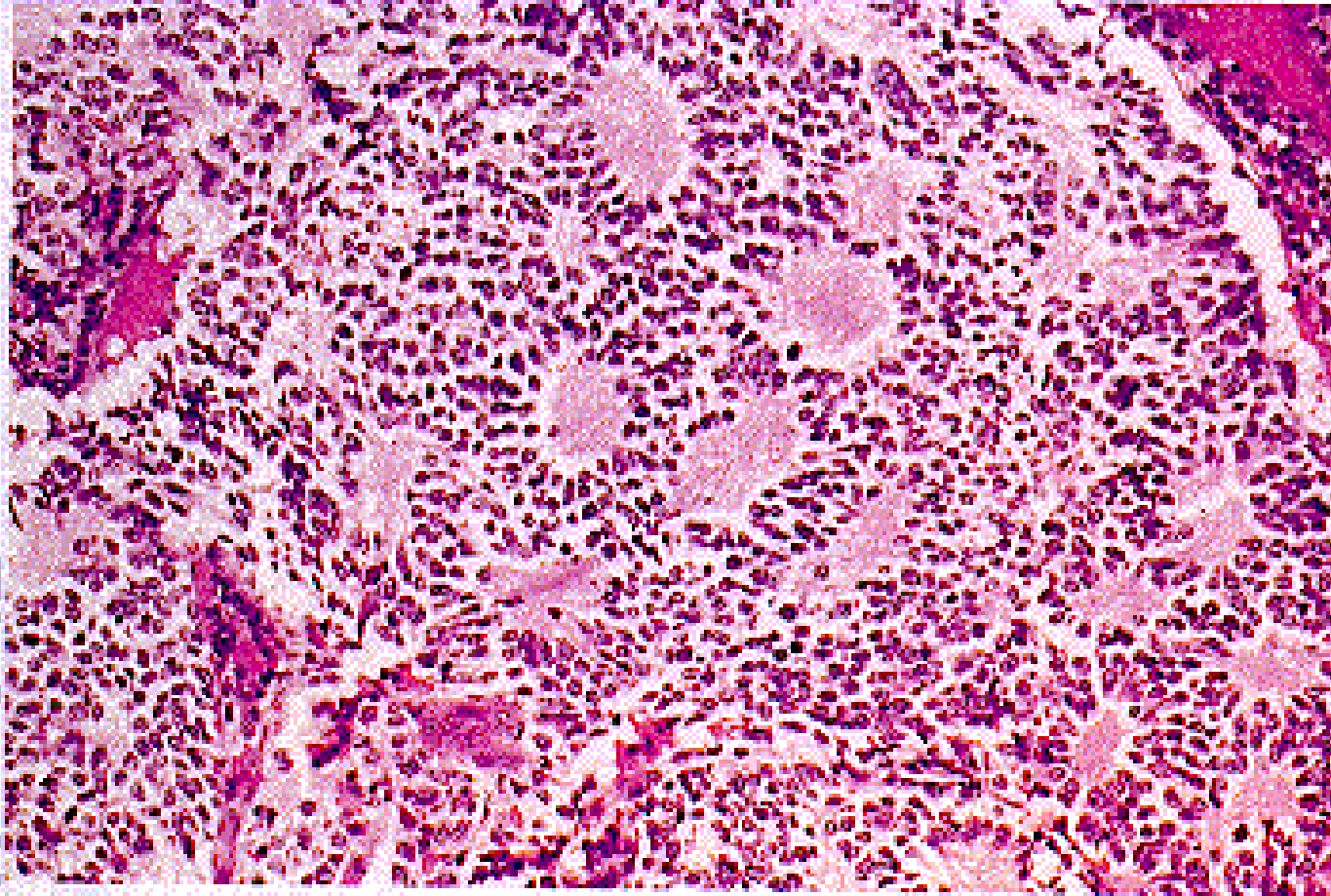
# Catecholamine Metabolism



Neuroblastoma can arise anywhere in the sympathetic nervous system. Primaries most commonly found in adrenal glands



# Typical histological appearance of neuroblastoma Showing rosettes of neoplastic cells



## Urine organic acids as an analytical tool

- The intermediary metabolites of most metabolic pathways are small organic molecules, amenable to detection by 'organic acid' analysis
- This is the most productive investigation in respect to detecting disturbances in metabolism, especially inborn
  - Will throw up surprises from time to time!



For more information or discussion about this case contact:

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