Knowledge of the natural history of paediatric MEN1 is required to inform decision making for predictive testing in childhood

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BACKGROUND

- Multiple Endocrine Neoplasia type 1 (MEN1), a dominantly inherited syndrome, is characterised by parathyroid hyperplasia, pancreatic neuroendocrine tumours (PNET) and pituitary adenomas, although more than 20 tumour types are described.
- Annual biochemical surveillance and abdominal imaging is recommended from <10yrs with pituitary imaging every 3yrs. [1]
- Age at start of surveillance is derived from the youngest reported patient with an MEN1 manifestation.

METHODS

Retrospective, observational study of paediatric patients screening according to international guidelines[1].

RESULTS

Patient	Age at diagnosis (years)	Number of hospital visits	No of blood tests (prolactin, IGF1, calcium and PTH)	No of MRIs (pituitary and abdominal)	Manifestations of MEN1
1	13	5	4	2	None
2	8	9	6	2	Asymptomatic eucalcaemic hyperparathyroidism
3	5	7	5	2	None
4	11	3	2	3	None
5	13	3	2	3	None
6	10	3	2	3	None
7	12	3	2	3	Asymptomatic eucalcaemic hyperparathyroidism
8	12	2	1	2	None
9	9	11	8	2	Asymptomatic eucalcaemic hyperparathyroidism
10	5	2	1	2	None
11	5	2	1	2	None
12	7	2	1	2	None
13	8	2	1	2	None

DISCUSSION

The evidence informing the management of paediatric patients with a change in the MEN1 gene is limited. The prevalence and natural history of tumours in childhood MEN1 is largely unknown. Surveillance imaging risks identification of incidental findings of uncertain significance, and the optimal timing of surgery for small, non-functioning PNETs and adrenal tumours is unknown.

Predictive testing is often undertaken in children too young to give consent and some adults, diagnosed in childhood, may have elected not to be tested.

n our small population, a large number of appointments, MRI scans and biochemical tests were performed and days at school and work were lost. To enable families to make informed decisions about predictive testing and surveillance, international collaboration is required to generate data describing the prevalence and natural history of MEN1 related tumours in childhood.

References
Thakker R et al. Clinical Practice Guidelines for Multiple Endocrine Neoplasia Type 1 (MEN1), *JCEM* (2012) 97: 9 (1); 2990–3011

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