

# The Involvement of Primary Care in the Management of Rare Disease in Ireland

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## Background/ Methods

This study involved a four year retrospective audit of ten Irish Primary Care practices (General Practitioners (GPs)), to examine their involvement in the diagnosis and management of rare disease patients.

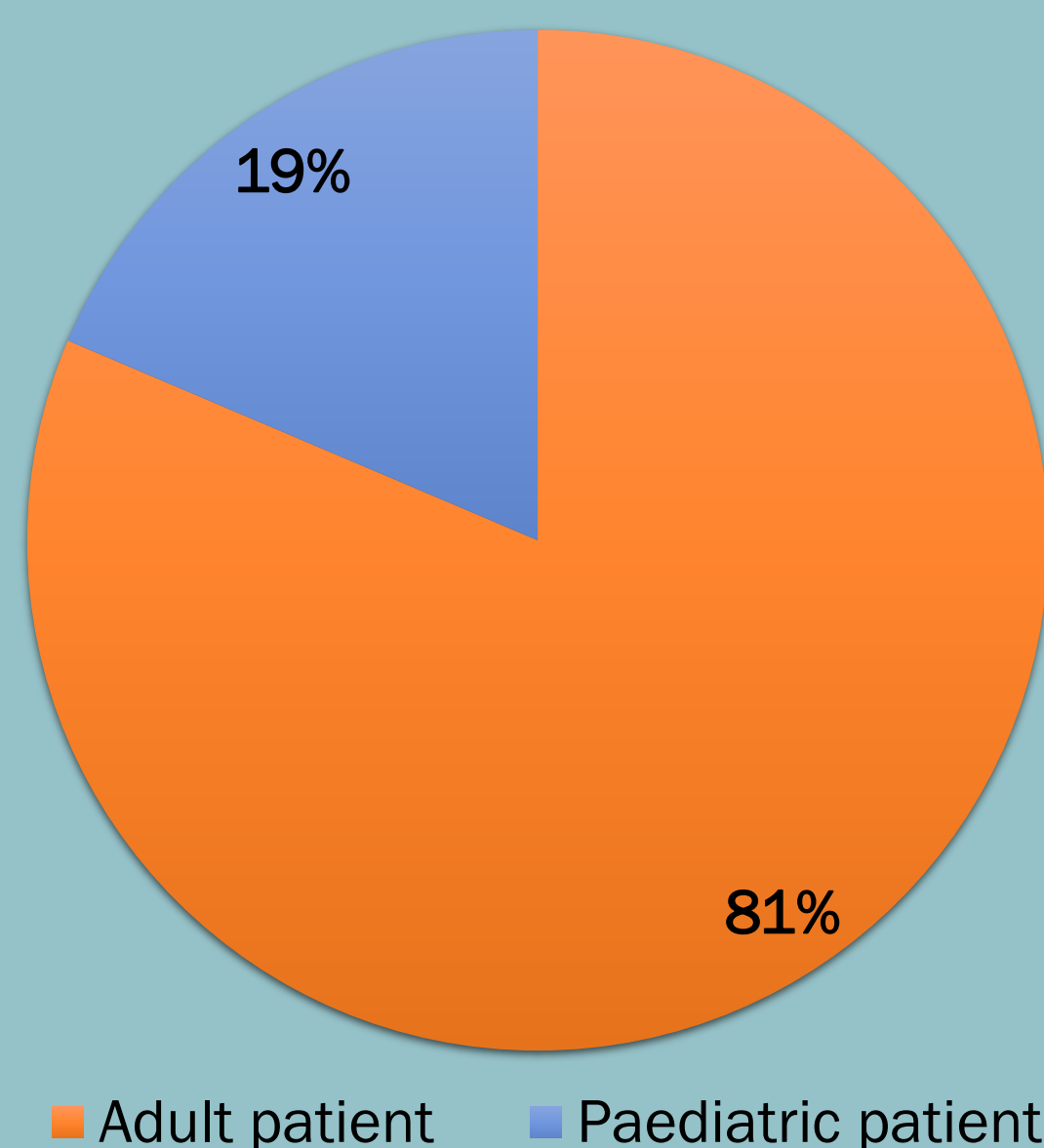
## Results

Overall 174 rare disease patients were identified. This does not correlate with the expected numbers of rare disease patients

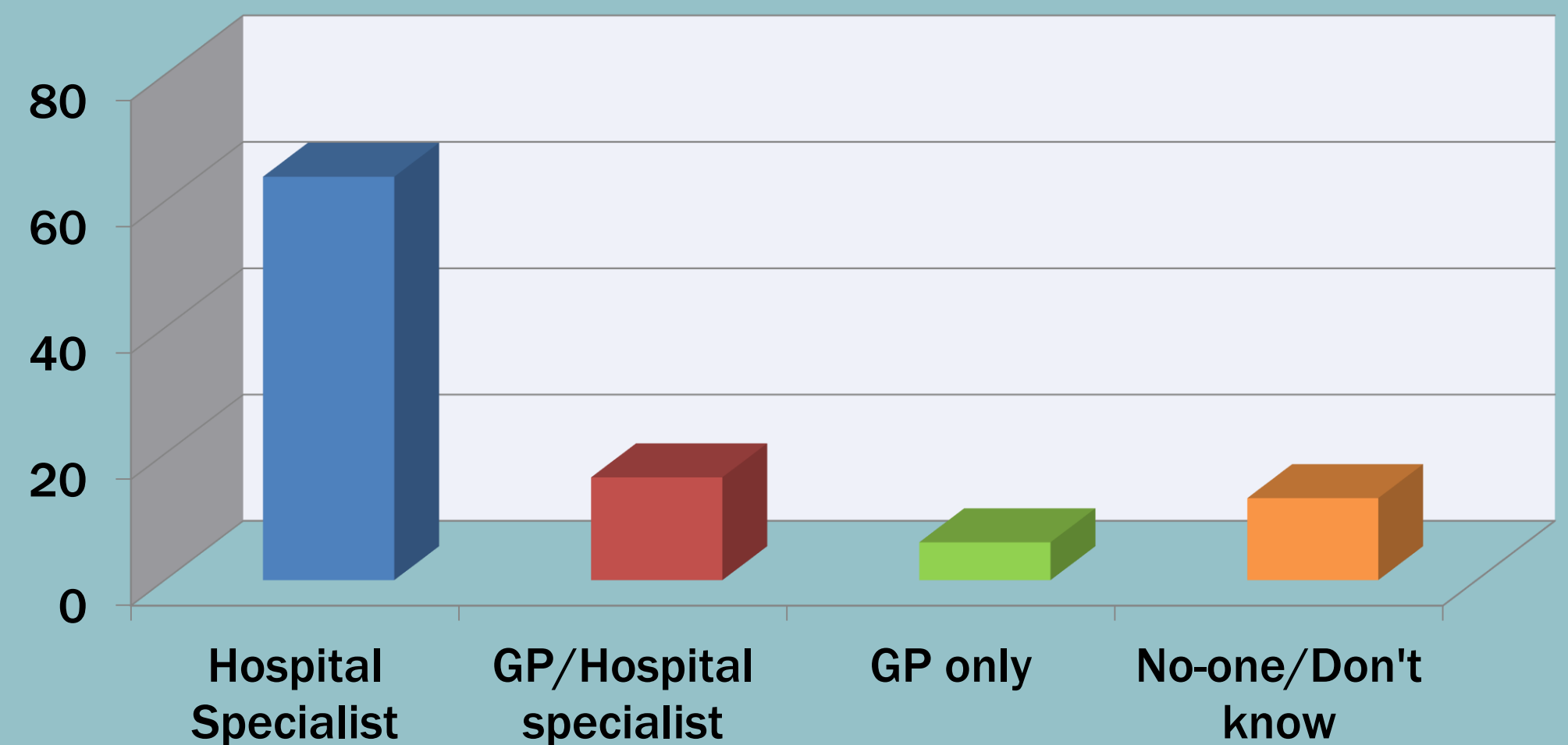
The numbers per practice were not comparable with the size of the practice

IT systems used by primary care teams are not equipped to identify rare disease patients. Specific coding for rare disease is not used

Coordination of care for children is facilitated by Paediatricians. Coordination of care for adult patients is generally GP based



**Figure 1. Proportion of Paediatric and Adult rare disease patients.** The audit revealed a disproportionate number of adult to paediatric rare disease patients (50% of those affected by rare disease are children (EURORDIS))



**Figure 2. Physicians managing the patients' rare disease**

## Discussion

Management of rare disease in Ireland is hospital based with little co-ordination of care at Primary Care level. This is inadequate for many patients.

This is at variance with other EU countries where care is mainly provided at primary care level, which would meet Irish patients' surveyed needs<sup>1,2</sup>. Rare disease patients who were eligible for state based GP care (GP visit/Medical card holders) had nearly twice the number of consultations (27.77/time period) than those that paid privately (14.6 consultations/time period).

These findings suggest that socioeconomic status influences attendance rates and the burden of disease.

## Conclusions

- Specific coding systems (e.g. ORPHA codes) are essential in terms of identifying rare diseases in Primary Care in Ireland.
- Co-ordination of care for rare disease patients at primary care level requires improvement in integrated care.
- Management of rare disease in Ireland is mainly by hospital specialists, unlike many other EU countries.
- Access to shared patient medical or electronic health record would improve communication, enhance patient safety and allow greater management and co-ordination of care for rare disease patients.

## References

1. Van Loenen, T et al. (2016) *BMC Fam Pract.* 17:59
2. Health Service Executive *Have your Say Public Consultation 2012*