Real-world Disease burden of Fibrodysplasia Ossificans Progressiva in China

RWD97

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BACKGROUND

Fibrodysplasia ossificans progressiva (FOP) is an ultra-rare, disabling genetic disorder that lacks real-world evidence on disease burden. Until 2018 fewer than 150 patients were identified in Mainland China. There's a serious lack of objective understanding of the patient's survival status.

OBJECTIVES

The study aims to investigate the basic characteristics, diagnostic status, prognosis, and disease burden of FOP patients in China.

METHODS

- ✓ An online survey was conducted through "China Cloud Platform for Rare Diseases" among patients recruited from the FOP patient organization in March 2023.
- ✓ Patient demographics characteristics, diagnosis, prognosis, healthcare resource usage and costs, and health-related quality of life data were collected.
- ✓ Health-related quality of life was assessed using EQ-5D-Y for children aged 4–15 and EQ-5D-5L for patients aged ≥ 16. Age subgroup analyses for those aged < 8, 8–15, and ≥ 16 were conducted.

RESULTS

✓ **Basic characteristics, diagnosis status and prognosis**

- ➢ A total of 67 patients (mean age **16.6±10.2** years, 43.3% female) were included. **62.7%** of patients were absent from school or unemployed due to FOP. **(Table 1)**
- ➢ The average delay of confirmed diagnosis was 3.1±4.3 years. Up to **78.8%** of patients would have been misdiagnosed at least once. As high as **97.0%** total population had heterotopic osteogenesis (HO) , **98.5%** of patients were disabled due to FOP. **(Table 1)**
- ➢ All subgroups showed high misdiagnosed rates and long delayed diagnosis duration among FOP patients, with patients aged ≥ 16 having the longest delayed diagnosis duration and the highest level of disability. **(Table 1)**

Table 1 Basic characteristics, diagnostic status and prognosis of FOP patients

Variables	Total population N=67	Age 0-8 N=18	Age 8-15 N=17	Age≥16 N=32
Basic characteristics				
Females,n (%)	29 (43.3%)	5 (27.8%)	9 (52.9%)	15 (46.9%)
Age,years,mean (SD)	16.6 (10.2)	5.7 (1.2)	11.5 (1.9)	25.5 (7.4)
Impact on Education or Employment, n (%)	42 (62.7%)	9 (50.0%)	4 (23.5%)	29 (90.6%)
Suspend or absent from school	29 (43.3%)	9 (50.0%)	4 (23.5%)	16 (50.0%)
Unemployment	13 (19.4%)	0 (0.0%)	0 (0.0%)	13 (40.6%)
Diagnostic status and prognosis				
Delayed diagnosis duration, years, mean, (SD)	3.1 (4.3)	0.7 (0.9)	1.1 (1.2)	5.5 (5.2)
Misdiagnosed patients, n (%)	52 (78.8%)	18 (100.0%)	13 (76.5%)	21 (65.6%)
Heterotopic Ossification, n (%)	65 (97.0%)	18 (100.0%)	16 (94.1%)	31(96.9%)
Disabled due to FOP, n (%)	66 (98.5%)	17 (94.4%)	17 (100.0%)	32 (100.0%)

✓ **Healthcare resource usuage and costs**

- ➢ Only **38.8%** of the patients had outpatient or inpatient visits in the past year, with 14.9% having hospitalisation (mean **1.8±1.6**) and 34.3% having outpatient visits (mean **2.6±1.9**), respectively. The healthcare resource utilization gradually decreases with age **(Figure 1).**
- ➢ The annual cost per patient was **¥74,250±80,105**, with **74.4%** being indirect costs in the total population. The highest total annual costs were for patients aged <8 years (¥97,969±105,894), followed by patients aged 8-15, and ≥16 years (¥58,623±61,134 & ¥69,210±59,641 respectively). **(Figure 2)**

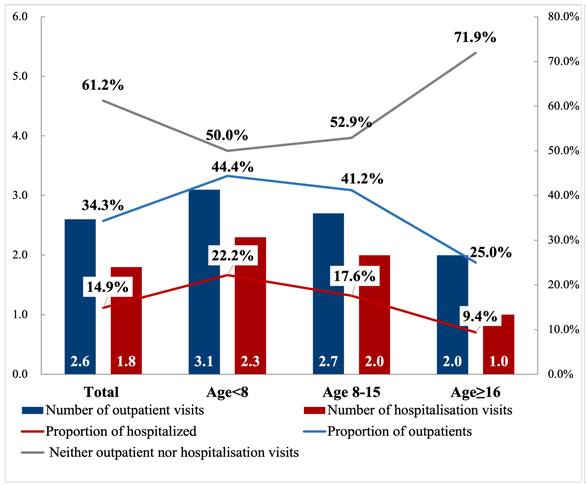

Figure 1 Annual healthcare resource utilization of FOP patients

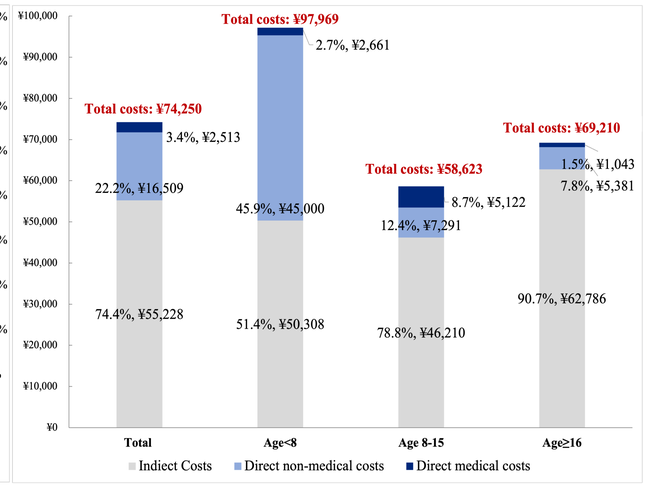

Figure 2 Annual costs and cost structure of FOP patients

✓ **Quality of life**

- ➢ Health utility values were lowest for patients aged ≥16 (**0.221±0.336**), compared to those aged <8 (**0.700±0.163**) and those aged 8–15 (**0.618±0.202**).

✓ **Quality of life**

- ➢ The dimension of **self-care** exhibited the most pronounced impairment in the overall quality of life among patients, affecting 86.7%, 100%, and 96.9% of patients in aged 8, 8–15, and ≥16 years subgroups, respectively.

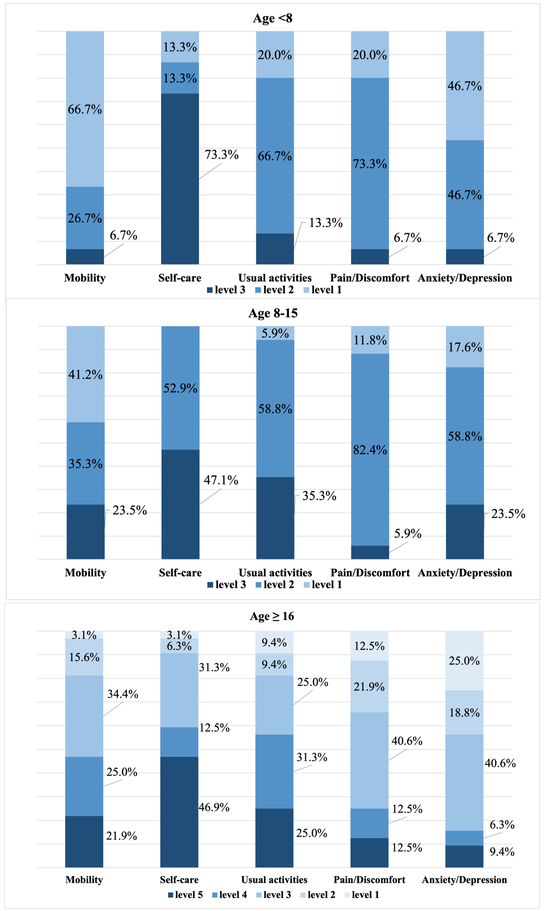

Figure 3 Distribution across levels of the EQ-5D dimensions of < 8, 8–15 and ≥16 years patients

CONCLUSIONS

FOP patients suffer high misdiagnosis rates, high disability rates, and inadequate disease management in China. High indirect costs and declining quality of life drove the significant disease burden.

Reference

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