### INTRODUCTION1,2

Cytochrome P450 oxidoreductase deficiency (PORD), which is caused by PORD gene variants, shows broad clinical features including skeletal malformations, steroidogenic defects, and disorders of sex development (DSD).

### AIM

- Genetic comprehensive analysis of PORD was reported in Japan1, however, clinical symptoms at diagnosis and the clinical course of PORD have not been reported.
- To reveal clinical symptoms at diagnosis and the clinical course of PORD in Japan.

### METHODS3

1. Questionnaire in Sep 2018: 65% response rate
   - 183 of JSPE councilors answered: 119 councilors answered
   - 39 PORD patients were examined at 20 hospitals

2. Questionnaire: 77% response rate
   - 39 examined PORD patients at 20 hospitals
   - Q: clinical features at diagnosis and clinical courses
   - 30 examined PORD patients (M:F = 13:17) at 18 hospitals

### RESULTS3

1. Age, height and BMI
   - At investigation: 12.1 (10.4–14.4) / 0.75 (0.73–0.8) / 95.9 (85.4–110.8)
   - At diagnosis: 0.21 (0.21–0.37)

2. Diagnosis determining symptoms

3. Urine steroid profile
   - 20 PORD patients were performed.
   - At 32 PORD patients were suspected with PORD.

4. External genitalia at diagnosis

5. Plastic surgery for external genitalia and puberty

6. PORD genetic analysis

7. Imaging tests

8. Development

### CONCLUSIONS3

- Urine steroid profile is useful to diagnose PORD.
- Japanese cases had many Amb574Ins variation as previously reported.
- External genitalia in females may be more severe than in males, and sex hormone replacement in females was the majority in pubertal stage.
- Hydrocortisone was major for steroid therapy.
- Administration methods are controversial as a permanent or only stress.

### REFERENCES


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