

**DISEASE:**  
**Progressive familial intrahepatic cholestasis type 5**

|                    |  |
|--------------------|--|
| <b>NAME:</b>       | Progressive familial intrahepatic cholestasis type 5                       |
| <b>ORPHACODE:</b>  | 480476   |
| <b>SYNONYMS:</b>   | NR1H4 deficiency<br>PFIC5  |
| <b>XREF(S):</b>    | <a href="#">Orphanet</a><br><a href="#">ICD-10</a><br><a href="#">OMIM</a> |
| <b>ANALYTE(S):</b> | <a href="#">NR1H4</a>  |
| <b>CREATED:</b>    | 13 May 2019 - 01:02  |
| <b>CHANGED:</b>    | 22 Jun 2023 - 16:14  |

---

Source URL: <http://gentest.healthdata.be/disease/2710>

## RELATED CONTENT

---

### Related Genetic Tests

- [Cholestasis \(gene panel\)](#)
- [Cholestasis, progressive familial intrahepatic \(gene panel\)](#)

### Related Laboratories

- [Centre de Génétique Médicale UCL](#)

### Related Analytes

- [nuclear receptor subfamily 1 group H member 4](#)

### Related Gene Panels

- [Cholestasis \(40 genes\) - UCL](#)

---

Source URL: <http://gentest.healthdata.be/disease/2710>