

**ANALYTE:**  
**PITX1**

|                             |                                                                                                                           |
|-----------------------------|---------------------------------------------------------------------------------------------------------------------------|
| <b>NAME:</b>                | paired like homeodomain 1                                                                                                 |
| <b>SYMBOL:</b>              | PITX1                                                                                                                     |
| <b>VERSION OF ORPHANET:</b> | 2023-06-22 14:14:43                                                                                                       |
| <b>SYNONYMS:</b>            | POTX<br>PTX1                                                                                                              |
| <b>XREF(S):</b>             | <u>Orphanet</u><br><u>Reactome</u><br><u>Ensembl</u><br><u>Genatlas</u><br><u>HGNC</u><br><u>OMIM</u><br><u>SwissProt</u> |
| <b>CREATED:</b>             | 13 May 2019 - 01:01                                                                                                       |
| <b>CHANGED:</b>             | 22 Jun 2023 - 16:14                                                                                                       |

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### Related Genetic Tests

- [Congenital malformation \(gene panel - 1721 genes\)](#)
- [Congenital malformation gene panel](#)
- [Skeletal dysplasia \(gene panel\)](#)
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- [Skeletal dysplasia \(gene panel\)](#)

### Related Diseases

- [Brachydactyly-elbow wrist dysplasia syndrome](#)
- [Familial clubfoot due to 5q31 microdeletion](#)
- [Familial clubfoot due to PITX1 point mutation](#)
- [Mirror-image polydactyly](#)

### Related Gene Panels

- [Congenital malformation \(1721 genes\) - ULB](#)
- [Congenital malformation gene panel - VUB](#)
- [Skeletal dysplasia \(394 genes\) - VUB](#)
- [Skeletal dysplasia \(genepanel\) - UZA](#)
- [Skeletal dysplasia - UGent](#)

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