|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| NOM PRENOM DDN | **Lieu de naissance**  | **Poids (Kg)** |  | **MS** |
| **Taille (cm)** |  |

|  |  |  |
| --- | --- | --- |
| Synthese et Descriptif**\*** |  |  |
| **Cranio****facial** | Dysmorphie faciale (3/5) : non [ ]  oui [ ]  | [ ]  **dolichocéphalie** [HP:0000268](https://hpo.jax.org/app/browse/term/HP%3A0000268)[ ]  **enophtalmie** [HP:0000490](https://hpo.jax.org/app/browse/term/HP%3A0000490)[ ]  **fentes palpébrales obliques vers le bas** [HP:0000494](https://hpo.jax.org/app/browse/term/HP%3A0000494) [ ]  **hypoplasie malaire** [HP:0012369](https://hpo.jax.org/app/browse/term/HP%3A0012369) [ ]  **rétrognathisme** [HP:0000278](https://hpo.jax.org/app/browse/term/HP%3A0000278) | m | 1 |
| Palais ogival [HP:0002705](https://hpo.jax.org/app/browse/term/HP%3A0002705) | [ ] **Absent** [ ] **Avec chevauchement dentaire**  [ ]  **Inconnu**[ ] **Sans chevauchement dentaire** | m |  |
| Hypertélorisme [HP:0000316](https://hpo.jax.org/app/browse/term/HP%3A0000316) | [ ] **Absent** [ ] **Présent** [ ] **Douteux** [ ]  **Inconnu** |  |  |
| Luette bifide [HP:0000193](https://hpo.jax.org/app/browse/term/HP%3A0000193) | [ ] **Absent** [ ] **Présent** [ ] **Douteux** [ ]  **Inconnu** |  |  |
| **Squelette** | Arachnodactylie [HP:0001166](https://hpo.jax.org/app/browse/term/HP%3A0001166) | [ ] **Absent**  [ ] **Signe du** **poignet** [ ]  **Inconnu** [ ]  **Signe du pouce**  | M | 13 |
| Flessum (Extension des coudes <170°) | [ ] **Absent** [ ] **Présent** [ ]  **Inconnu** | M | 1 |
| Hypermobilité articulaire : non [ ]  oui [ ]  (score de Beigton : /9) [HP:0001382](https://hpo.jax.org/app/browse/term/HP%3A0001382) | **Pouce contre avant-bras Droit :** [ ]  **Gauche**[ ] **5ème Doigt 90° Droit :** [ ]  **Gauche** [ ]  **Mains-sol** [ ] **Récurvatum coude >10° Droit :** [ ]  **Gauche**[ ] **Récuvatum genou >10° Droit :** [ ]  **Gauche** [ ]  | m |  |
| Pectus  | [ ]  **Absent**  [ ]  **Carinatum** [HP:0000768](https://hpo.jax.org/app/browse/term/HP%3A0000768)[ ]  **Inconnu**[ ]  **Asymétrie** [HP:0000766](https://hpo.jax.org/app/browse/term/HP%3A0000766) [ ] **Excavatum** [HP:0000767](https://hpo.jax.org/app/browse/term/HP%3A0000767) | MM | 21 |
| Dolichosténomelie [HP:0001519](https://hpo.jax.org/app/browse/term/HP%3A0001519) (E/T> 1.05) | [ ] **Absent** [ ]  **Présent E/T =……….** [ ] **Inconnu** | M | 1 |
| Pied plat [HP:0001763](https://hpo.jax.org/app/browse/term/HP%3A0001763) | [ ] **Absent** [ ] **Sans déformation de l’arrière pied** [ ]  **Inconnu** [ ] **Avec déformation de l’arrière pied**  | M | 21 |
| Protrusion acétabulaire [HP:0003179](https://hpo.jax.org/app/browse/term/HP%3A0003179) | [ ] **Absent** [ ] **Présent** [ ] **Douteux** [ ]  **Inconnu** | M | 2 |
| Cyphose thoracolombaire [HP:0002808](https://hpo.jax.org/app/browse/term/HP%3A0002808) | [ ] **Absent** [ ] **Présent** [ ]  **Inconnu** |  | 1 |
| Scoliose > 20° [HP:0002650](https://hpo.jax.org/app/browse/term/HP%3A0002650) | [ ] **Absent** [ ] **Présent** [ ]  **Inconnu** | M |
| Spondylolisthésis [HP:0003302](https://hpo.jax.org/app/browse/term/HP%3A0003302) | [ ] **Absent** [ ] **Présent** [ ]  **Inconnu** |  |
| **Pulmonaire** | Pneumothorax spontané [HP:0002108](https://hpo.jax.org/app/browse/term/HP%3A0002108) | [ ] **Absent** [ ] **Présent** [ ]  **Inconnu** | m | 2 |
| Bulle apicale [HP:0032176](https://hpo.jax.org/app/browse/term/HP%3A0032176) | [ ] **Absent** [ ] **Présent** [ ]  **Inconnu** | m |  |
| **Cutané** | Hernie récidivante [HP:0000023](https://hpo.jax.org/app/browse/term/HP%3A0000023) | [ ] **Absent** [ ] **Présent** [ ]  **Inconnu** | m |  |
| Vergetures [HP:0001065](https://hpo.jax.org/app/browse/term/HP%3A0001065) | [ ] **Absent** [ ] **Présent** [ ]  **Non significatif** [ ]  **Inconnu** [ ] **Hanche** [ ] **Seins** [ ] **Épaules** [ ] **Dorsales** [ ] **Fesses** [ ] **Cuisses** [ ] **Ventre**[ ] **Lombaires** | m | 1 |
| Livedo reticularis [HP:0000965](https://hpo.jax.org/app/browse/term/HP%3A0000965) | [ ] **Absent** [ ] **Présent** [ ] **Douteux** [ ]  **Inconnu** |  |  |
| Hyperlaxité cutanée [HP:0001388](https://hpo.jax.org/app/browse/term/HP%3A0001388) | [ ] **Absent** [ ] **Présent** [ ] **Douteux** [ ]  **Inconnu** |  |  |
| **Neuro** | Ectasie durale lombo-sacrée [HP:0100775](https://hpo.jax.org/app/browse/term/HP%3A0100775) | [ ] **Absent** [ ] **Présent** [ ]  **Inconnu** | M | 2 |
| Retard des apprentissages [HP:0001328](https://hpo.jax.org/app/browse/term/HP%3A0001328) | [ ] **Absent** [ ] **Présent** [ ]  **Inconnu** |  |  |
| Antécédents d’épilepsie [HP:0001250](https://hpo.jax.org/app/browse/term/HP%3A0001250) | [ ] **Absent** [ ] **Présent** [ ]  **Inconnu** |  |  |
| **Oculaire** | Ectopie du cristallin [HP:0001083](https://hpo.jax.org/app/browse/term/HP%3A0001083) | [ ] **Absent** [ ] **Douteux** [ ] **Présent**  [ ]  **Inconnu** [ ]  **Aphake** [ ]  **Pseudo aphake** | M | 0 |
| Myopie > 3 dioptries [HP:0000545](https://hpo.jax.org/app/browse/term/HP%3A0000545) | [ ] **Absent** [ ] **Présent** [ ]  **Inconnu** | m | 1 |
| Cornée plate [HP:0007720](https://hpo.jax.org/app/browse/term/HP%3A0007720) | [ ] **Absent** [ ] **Présent** [ ]  **Inconnu** | m |  |
| Iris Floculi [HP:0500007](https://hpo.jax.org/app/browse/term/HP%3A0500007) | [ ] **Absent** [ ] **Présent** [ ]  **Inconnu** |  |  |
| **Cardio-****Vasculaire** | Dissection de l’aorte  | [ ] **Absent** [ ]  **Type A** [HP:0004933](https://hpo.jax.org/app/browse/term/HP%3A0004933)[ ]  **Type B** [HP:0012499](https://hpo.jax.org/app/browse/term/HP%3A0012499) **Age :** …………… [ ]  **Inconnu** | M | 0 |
| Chirurgie de l’aorte ascendante | [ ] **Absent** [ ] **Présent Age :** …………................[ ]  **Inconnu** |
| TAA [HP:0004970](https://hpo.jax.org/app/browse/term/HP%3A0004970)  | Sinus de Valsalva | ……..….mm z-score : |
| Aorte tubulaire | …………mm z-score : |  |  |
| Dilatation artère pulmonaire [HP:0004927](https://hpo.jax.org/app/browse/term/HP%3A0004927) | [ ] **Absent** [ ] **Présent** [ ]  **Inconnu** | m |  |
| Aorte abdominale  | [ ] **Absent** [ ]  **dissection** [ ]  **dilatation** [HP:0002636](https://hpo.jax.org/app/browse/term/HP%3A0002636) **Age :….**  | m |  |
| Facteurs de risque | [ ] **Absent** [ ]  **HTA** [HP:0000822](https://hpo.jax.org/app/browse/term/HP%3A0000822)[ ]  **Inconnu** [ ] **Autre** : ……… |  |  |
| Valve aortique | [ ]  **Tricuspide** [ ]  **Bicuspide** [HP:0001647](https://hpo.jax.org/app/browse/term/HP%3A0001647)[ ]  **Inconnu** |  |  |
| Mitrale | [ ]  **Normale** [ ]  **Prolapsus** [HP:0001634](https://hpo.jax.org/app/browse/term/HP%3A0001634)[ ]  **Inconnu** [ ]  **Age de la chirurgie**:…… | m | 1 |
| Tortuosité artérielle [HP:0006687](https://hpo.jax.org/app/browse/term/HP%3A0006687) | [ ] **Absent** [ ] **Présent** [ ] **Douteux** [ ]  **Inconnu** | Total :  |
| Cardiopathie congénitale | [ ] **Absent** [ ] **PCA** [HP:0001643](https://hpo.jax.org/app/browse/term/HP%3A0001643) [ ] **CIV** [HP:0001629](https://hpo.jax.org/app/browse/term/HP%3A0001629)[ ]  **CIA** [HP:0001631](https://hpo.jax.org/app/browse/term/HP%3A0001631)  |
| **Génétique et Arbre G** | [ ]  Arbre généalogique transmis | [ ]  Nimblegen MFS-TAA [ ]  TSCA design [ ]  Caryotype [ ] Autre :………………………. |
| **Commentaire** |  |
| **Avis de la RCP** |  |

SYNTHESE DU