

GPS 新病例创建 使用手册

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Step1:

点击网页进入系统首页：<http://www.diseasegps.org>

The screenshot shows the homepage of the DiseaseGPS system. At the top, there are social media icons (Facebook, Twitter, LinkedIn, Instagram, YouTube) and a navigation bar with links for "登录/注册" (Login/Register), "English", and "常见问题" (FAQ). Below the navigation bar is the "DiseaseGPS phenotype search" logo and several institutional logos. The main content area features a navigation menu with tabs for "症状诊断" (Symptom Diagnosis), "基因诊断" (Gene Diagnosis), "综合诊断" (Comprehensive Diagnosis), "病例库" (Case Library), "VCF SERVER", and "关于我们" (About Us). The "症状诊断" tab is active, displaying a "隐藏症状树" (Hidden Symptom Tree) on the left. The tree is expanded to show categories like "遗传模式" (Inheritance Mode), "表型异常" (Phenotypic Abnormalities), "泌尿生殖系统异常" (Urogenital System Abnormalities), "头部和颈部的异常" (Head and Neck Abnormalities), "眼部异常" (Eye Abnormalities), "耳部异常" (Ear Abnormalities), "神经系统异常" (Neurological Abnormalities), "乳房异常" (Breast Abnormalities), "内分泌系统异常" (Endocrine System Abnormalities), "骨骼系统的异常" (Skeletal System Abnormalities), "胎儿产前发育或出生异常" (Fetal Prenatal Development or Birth Abnormalities), "生长异常" (Growth Abnormalities), "体壁的异常" (Body Wall Abnormalities), and "声音异常" (Voice Abnormalities). On the right, there is a search bar with the placeholder text "请输入症状名或症状ID" (Please enter symptom name or symptom ID) and a "搜索" (Search) button. Below the search bar, there is a recommendation section with "眼部异常" (Eye Abnormalities), "耳部异常" (Ear Abnormalities), and "神经系统异常" (Neurological Abnormalities) highlighted. A message below the search bar reads: "请在搜索栏中输入，或点击左侧树状图中的箭头按钮，即可显示详细信息" (Please enter in the search bar, or click the arrow button in the tree diagram on the left, to display detailed information).

Step2:

点击登陆/注册按钮进行用户登陆或者注册

This screenshot is identical to the one above, showing the DiseaseGPS homepage. The key difference is that the "登录/注册" (Login/Register) button in the top navigation bar is highlighted with a red rectangular box, indicating the step to click for user login or registration.



Step3: 进入用户界面，点击“新建病例”进行病例添加



Step4: 输入依次新病例的表型，基因型等信息，并确认提交

新建病历

浏览权限* 私有的

表型异常*
 ×语言发育迟缓 ×言语不能 ×中度智力残疾
 ×社交障碍 ×运动发育迟缓 ×眉毛稀疏
 ×下眼睑浮肿 ×鼻梁凹陷 ×鼻翼扁平
 ×薄嘴唇

来源医院* 上海市儿童医院

患者名字

性别* 男

患者年龄

备注
 语言发育迟缓, 言语不能, 中度智力残疾, 社交障碍, 运动发育迟缓, 眉毛稀疏, 下眼睑浮肿, 鼻梁凹陷, 鼻翼扁平, 薄嘴唇

相关疾病
 请输入最终诊断的OMIM号

提交 返回

Step5:

诊断结果如下，可选择症状诊断，基因诊断以及综合诊断，一般来说参考综合诊断结果。

admin | 退出登陆 | English | 常见问题

phenotype search

症状诊断 基因诊断 综合诊断 病例库 VCF SERVER 关于我们

Score: 0.68

1. CHARGE SYNDROME
 (分数: 0.68, P值: <0.1)

CHARGE syndrome is a congenital condition (present from birth) that affects many areas of the body. CHARGE stands for coloboma, heart defect, atresia choanae (also known as choanal atresia), restricted growth and development, genital abnormality, and ear abnormality.

同义词:
 CHARGE ASSOCIATION--COLOBOMA, HEART ANOMALY, CHOANAL ATRESIA, RETARDATION, GENITAL AND EAR ANOMALIES
 HALL-HITTNER SYNDROME; HHS

OMIM基因(染色体位置): CHD7(8q12.2); SEMA3E(7q21.11);
 HPO基因(染色体位置): CHD7(8q12.2); SEMA3E(7q21.11);
 遗传模式: 常染色体显性遗传;散发的;
 死亡年龄: -
 发病率: -
 INDEL: -
 临床调节因素: 表型多样性;
 发病年龄: 新生儿期发病;
OMIM: 214800